

NATIONAL HUMAN GENOME RESEARCH INSTITUTE Division of Intramural Research



2006

*Current Topics in Genome Analysis*  
*Fall 2006*

*Week 4: Mining Genomic Sequence Data*

*Tyra G. Wolfsberg, Ph.D.*

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES | NATIONAL INSTITUTES OF HEALTH | genome.gov DIR



Accessing public genome sequence data

UCSC's Genome Browser ("Golden Path")  
<http://genome.ucsc.edu>

NCBI's Map Viewer  
<http://www.ncbi.nlm.nih.gov/mapview/>

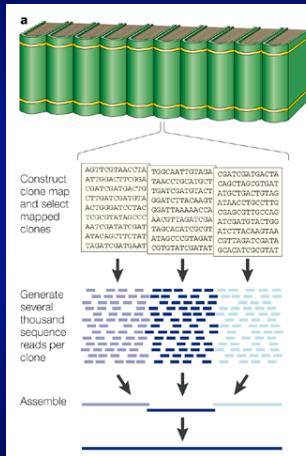
Ensembl  
<http://www.ensembl.org>

## Types of data integrated in genome browsers

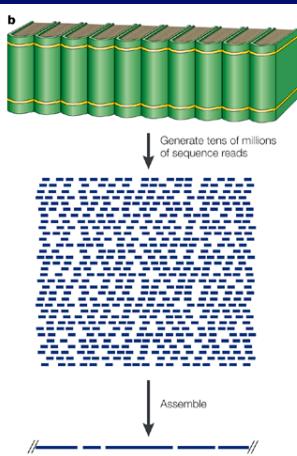
- Same starting material for all genome browsers: genomic sequence
- Annotations calculated independently by each genome browser
  - Genes
    - RefSeq mRNAs (non-redundant)
    - GenBank mRNAs (redundant)
    - ESTs
    - Gene predictions
  - SNPs
  - Homologous sequences from other organisms
  - STSs

## Overview of genome sequencing strategies

Clone-by-clone shotgun sequencing



Whole-genome shotgun sequencing



Nature Reviews | Genetics  
Green ED. Strategies for the systematic sequencing of complex genomes.  
Nat Rev Genet. 2001; 2:573-83.

## Genome Sequence Assemblies

- Complex algorithms needed to incorporate all sequence data
- Assemblies updated periodically as new sequence becomes available
  - Mouse and human genomes assembled by NCBI
  - Other genomes assembled by sequencing centers or consortia
- Assemblies not updated concurrently by the three Genome Browsers
  - “Pre-release” assemblies and annotations available at
    - UCSC: <http://genome-test.cse.ucsc.edu/>
    - pre!Ensembl: <http://pre.ensembl.org/>
  - UCSC and Ensembl provide access to older genome assemblies and annotations; NCBI provides access only to old mouse and human data
- IF YOU ARE COMPARING DATA FROM DIFFERENT GENOME BROWSERS, MAKE SURE YOU ARE LOOKING AT THE SAME VERSION OF THE ASSEMBLY

## Genome Assembly Versions

	Same assembly?	UCSC	NCBI	Ensembl
Human	Yes	Mar 2006/hg18/Build 36.1	Build 36.1	Build 36
Mouse	YES	Feb 2006/mm8/Build 36	Build 36.1	Build 36
Rat	YES	Nov 2004/rn4/RGSC 3.4	RGSC 3.4	RGSC 3.4
Zebrafish	NO	Mar 2006/danRer4/Zv6	Build 1.1/Zv4	Zv6
Rhesus	YES	Jan 2006/rheMac2/v.1.0, Mmul_051212	Build 1.1/v.1.0, Mmul_051212	Mmul_1
Fugu	NO	Aug 2002/ fr1/v3.0	-	Fugu 4.0

## NCBI Reference Sequences (RefSeqs)

- Derived from primary GenBank submissions
- Varying levels of validation, additional annotation, and manual curation

NC_123456	Genomic	Mixed	Complete genomic molecules including genomes, chromosomes, organelles, plasmids.	NT_123456	Genomic	Automated	Intermediate genomic assemblies of BAC and/or Whole Genome Shotgun sequence data
NG_123456	Genomic	Mixed	Incomplete genomic region supplied to support the NCBI Genome Annotation pipeline. Represents either non-transcribed pseudogenes, or larger regions representing a gene cluster that is difficult to annotate via automatic methods.	NW_123456	Genomic	Automated	Intermediate genomic assemblies of BAC or Whole Genome Shotgun sequence data
NM_123456	mRNA	Mixed	Transcript products; Mature RNA (mRNA) protein-coding transcripts.	NZ_ABCD12345678	Genomic	Automated	A collection of whole genome shotgun sequence data for a project. Accessions are not named for individual samples. The first four characters following the underscore (e.g., 'ABCD') identifies a genome project.
NM_123456789	mRNA	Mixed	Transcript products; 9-digit expansion of accession series	XM_123456	mRNA	Automated	Transcript products; model mRNA provided by the Genome Annotation process; sequence corresponds to the genomic contig.
NP_123456	Protein	Mixed	Protein products; primarily full-length precursor products but may include some partial proteins and mature peptide products.	XP_123456	Protein	Automated	Protein products; model proteins provided by the Genome Annotation process; sequence corresponds to the genomic contig.
NP_123456789	Protein	Curation	Protein products; 9-digit expansion of accession series	XR_123456	RNA	Automated	Transcript products; model non-coding transcripts provided by the Genome Annotation process.
NR_123456	RNA	Mixed	Non-coding transcripts including structural RNAs, transcribed pseudogenes, and others				

<http://www.ncbi.nlm.nih.gov/RefSeq/key.html>

### Beta actin mRNA RefSeq

LOCUS NM\_001101 1793 bp mRNA linear PRI 17-SEP-2006

DEFINITION Homo sapiens actin, beta (ACTB), mRNA.

ACCESSION NM\_001101

VERSION NM\_001101.2 GI:5016088

KEYWORDS .

SOURCE Homo sapiens (human)

ORGANISM

  Bukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhines 1 to 1793

REFERENCE 1 (bases 1 to 1793)

AUTHORS Pappenberg, G., McCormick, E.A. and Willison, K.R.

TITLE Quantitative actin folding reactions using yeast CCT purified via an in vitro assay in the Cdc48/p97 subunit

JOURNAL J. Mol. Biol. 360 (2), 484-496 (2006)

PUBMED 16762366

.....

REFERENCE 154 (bases 1 to 1793)

AUTHORS Vandervelde, J., Lapiere, C.M. and Weber, K.

TITLE Human cytoskeletal actins are the products of at least two genes and differ in primary structure in at least 25 identified positions from skeletal muscle actins

JOURNAL Proc. Natl. Acad. Sci. U.S.A. 75 (3), 1106-1110 (1978)

PUBMED 274701

COMMENT REFERENCE [REMOVED]. This record has been curated by NCBI staff. The reference sequence was derived from NM\_001101 and X61472. On Jun 8, 1999 this sequence version replaced gi:1501884.

Summary: Beta actin is one of six different actin isoforms which have been identified. ACTB is one of the two nonmuscle cytoskeletal actins. Actins are highly conserved proteins that are involved in maintaining cell motility and integrity. Alpha actins are a major constituent of the contractile apparatus.

COMPLETENESS: complete on the 3' end.

.....

CDS

74..1201

(gene="ACTB")

(product="beta actin")

(protein\_id="NP\_0010921")

(db\_xref="NC\_010921")

(db\_xref="CCDS00000341\_1")

(db\_xref="GeneID:60")

(db\_xref="HPID:1302")

(db\_xref="MI:1024")

(db\_xref="MIM:102430")

(transl\_table=11)

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VAKVYVLEKLGIVLAVVVPYRDLVLLVAVVAVYRRPGLGVVPCGGCCEHSD

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RKYWSWIGGSILASLSTFQTMQWISIKQEYDESCPSIVHRKCF"

.....

ORIGIN

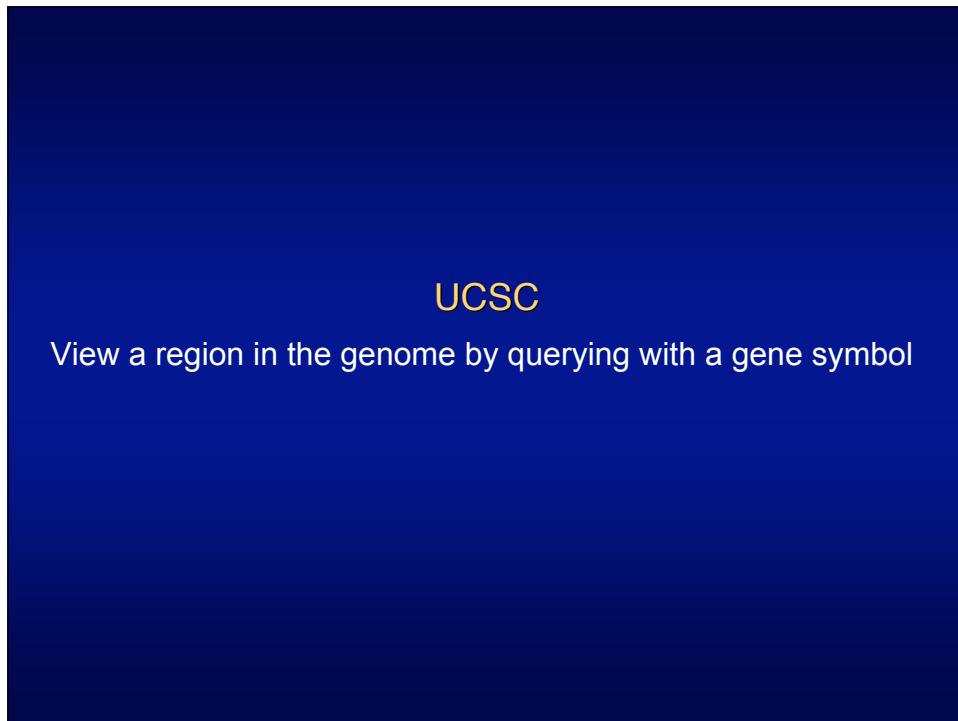
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121 qtcgggcgg gtcggcggcc gtcggatgc ccccccggcc gtcggccct ccggcgccg

181 ggccggccgg caccaggccg tgatgttgg catgggtccg aacgatccat atgtggccg

241 cggggccgg agcaagagag acatccatcc ctgtggatcc cccatggccg acggatcg

..



UCSC Genome Bioinformatics

Genomes - Blat - Tables - Gene Sorter - PCR - Proteome - FAQ - Help

**Genome Browser** (highlighted with a red box)

**About the UCSC Genome Bioinformatics Site**

This site contains the reference sequence and working draft assemblies for a large collection of genomes. It also provides a portal to the ENCODE project.

We encourage you to explore these sequences with our tools. The Genome Browser zooms and scrolls over chromosomes, showing the work of annotators worldwide. The Gene Sorter shows expression, homology and other information on groups of genes that can be related in many ways. Blat quickly maps your sequence to the genome. The Table Browser provides convenient access to the underlying database. VisiGene lets you browse through a large collection of *in situ* mouse and frog images to examine expression patterns.

**News**

To receive announcements of new genome assembly releases, new software features, updates and training seminars by email, subscribe to the [genome-announce](#) mailing list.

**8 September 2006 - Old Rhesus Assembly Archived**

The Jan. 2005 *rheMac1* draft assembly has been moved from the main Genome Browser website to our [archive server](#). The data remain available for browsing and downloading, although blat services are no longer supported.

**8 August 2006 - New Opossum Assembly Available in Genome Browser**

The UCSC Genome Brower now includes the latest draft assembly of the opossum genome. The Jan. 2006 release of *Monodelphis domestica* (UCSC version monDom4) was sequenced and assembled by [The Broad Institute](#), Cambridge, MA, USA.

This draft, which has approximately 6.5X coverage, has an assembly length of nearly 3.61 billion bp including gaps (3.50 billion bp without gaps) contained on chromosomes 1-8, X, and Un. The N50 of the genome including gaps is 104,359 bp; the N50 without gaps is 107,990. The N50 size is the length such that 50% of the assembled genome lies in blocks of the N50 size or longer.

The monDom4 sequence and annotation data can be downloaded from the Genome Brower [FTP server](#) or [Downloads](#) page. Please review the [guidelines](#) for using the opossum assembly data.

Many thanks to The Broad Institute for providing these data. The UCSC opossum Genome Brower was produced by Hiram Clawson, Archana Thakkapallayil, Ann Zweig, Kayla Smith and Donna Karolchik. The initial set of annotation tracks was generated by the UCSC Genome Bioinformatics Group. See the Genome Brower [Credits](#) page for a detailed list of the organizations and individuals who contributed to the release of this browser.

Human (Homo sapiens) Genome Browser Gateway

Home Genomes Blat Tables Gene Sorter PCR FAQ Help

Human (Homo sapiens) Genome Browser Gateway

The UCSC Genome Browser was created by the [Genome Bioinformatics Group of UC Santa Cruz](#). Software Copyright (c) The Regents of the University of California. All rights reserved.

clade	genome	assembly	position or search term	image width
Vertebrate	Human	Mar. 2006	ADAM2	620
<input type="button" value="reset"/> the browser user interface settings to their defaults. <input type="button" value="tracks"/> <input type="button" value="configure tracks and display"/> <input type="button" value="clear position"/>				

**About the Human Mar. 2006 (hg18) genome**

The March 2006 human reference sequence (X. tropicalis) was produced by the International Human Genome Sequencing Consortium.

**Sample position queries**

A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS marker, or a cytological band, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of valid position queries for the human genome. See the [User's Guide](#) for more information.

**Request:** Genome Brower Response:

chr7 Displays all of chromosome 7  
 20p13 Displays region for band p13 on chr 20  
 chr3:1-1000000 Displays first million bases of chr 3, counting from p arm telomere  
 chr3:1000000+2000 Displays a region of chr3 that spans 2000 bases, starting with position 1000000

D16S3046 Displays region around STS marker D16S3046 from the Genethon/Marshfield maps. Includes 100,000 bases on each side as well.  
 RH18061:RH80175 Displays region between STS markers RH18061:RH80175. Includes 100,000 bases on each side as well. This syntax may also be used for other range queries, such as between cytobands and uniquely-determined ESTs, mRNAs, refSeqs, etc.

AA205474 Displays region of EST with GenBank accession AA205474 in BRCA1 cancer gene on chr 17  
 AC008101 Displays region of clone with GenBank accession AC008101  
 AF083811 Displays region of mRNA with GenBank accession number AF083811  
 PRNP Displays region of genome with HUGO Gene Nomenclature Committee identifier PRNP  
 NM\_017414 Displays the region of genome with RefSeq identifier NM\_017414  
 NP\_059110 Displays the region of genome with protein accession number NP\_059110

pseudogene mRNA Lists transcribed pseudogenes, but not cDNAs  
 homeobox caudal Lists mRNAs for caudal homeobox genes  
 zinc finger Lists many zinc finger mRNAs

[Done](#)

Human ADAM2 – UCSC Genome Browser v141

Known Genes

ADAM2 (NM\_001464) at chr8:39720414-39814936 - ADAM metallopeptidase domain 2 proprotein  
 ADAM2 (BC039899) at chr8:39720414-39814936 - ADAM metallopeptidase domain 2 proprotein  
 ADAM2 (BC034957) at chr8:39720414-39814936 - ADAM protein.  
 ADAM22 (NM\_021723) at chr7:87401638-87649383 - ADAM metallopeptidase domain 22 isoform 1  
 ADAM22 (NM\_021721) at chr7:87401638-87649364 - ADAM metallopeptidase domain 22 isoform 5  
 ADAM22 (NM\_016351) at chr7:87401638-87649383 - ADAM metallopeptidase domain 22 isoform 3  
 ADAM22 (NM\_021722) at chr7:87401638-87649305 - ADAM metallopeptidase domain 28 preproprotein  
 ADAM28 (NM\_014265) at chr8:24207565-24268659 - ADAM metallopeptidase domain 28 isoform 1  
 ADAM28 (NM\_004194) at chr7:87401638-87649364 - ADAM metallopeptidase domain 22 isoform 4  
 ADAM28 (NM\_003814) at chr14:70058832-70071486 - ADAM metallopeptidase domain 20 preprotein  
 ADAM28 (NM\_003813) at chr14:70058832-70064952 - ADAM metallopeptidase domain 21 preprotein  
 ADAM28 (NM\_003813) at chr14:70058832-70064952 - ADAM metallopeptidase domain 23 preprotein  
 ADAM20 (AP029899) at chr14:70058832-70061255 - ADAM metallopeptidase domain 20  
 ADAM22 (BC036029) at chr7:87401502-87600049 - ADAM22 protein.  
 ADAM22 (AF155381) at chr7:87401671-87663791 - Hypothetical protein ADAM22 (Fragment).  
 ADAM18 (AY358321) at chr8:13956125-39706780 - ADAM metallopeptidase domain 18

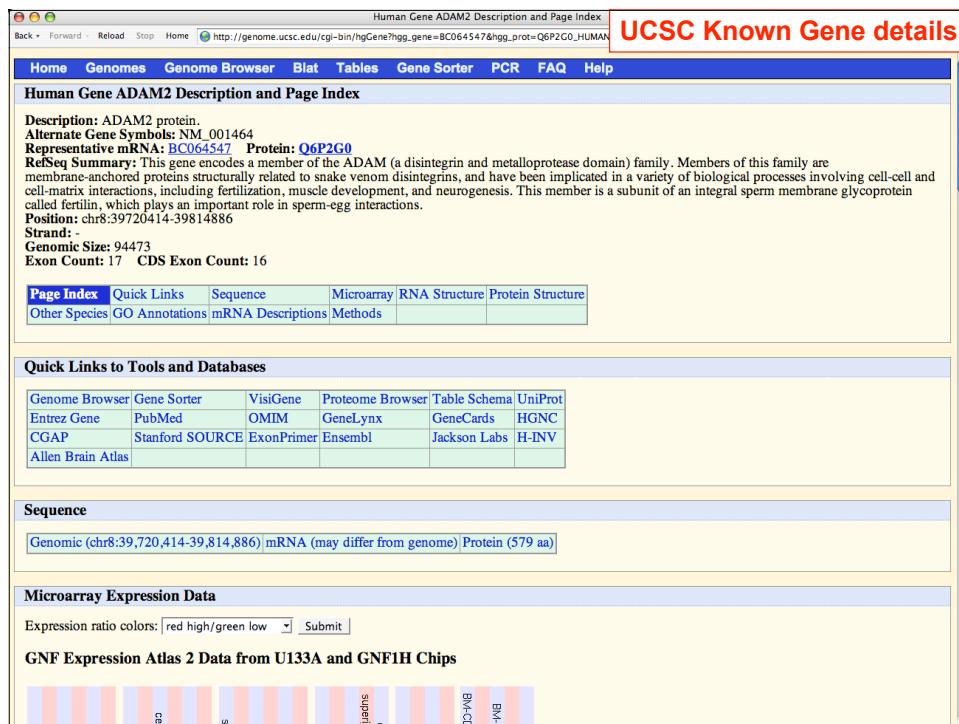
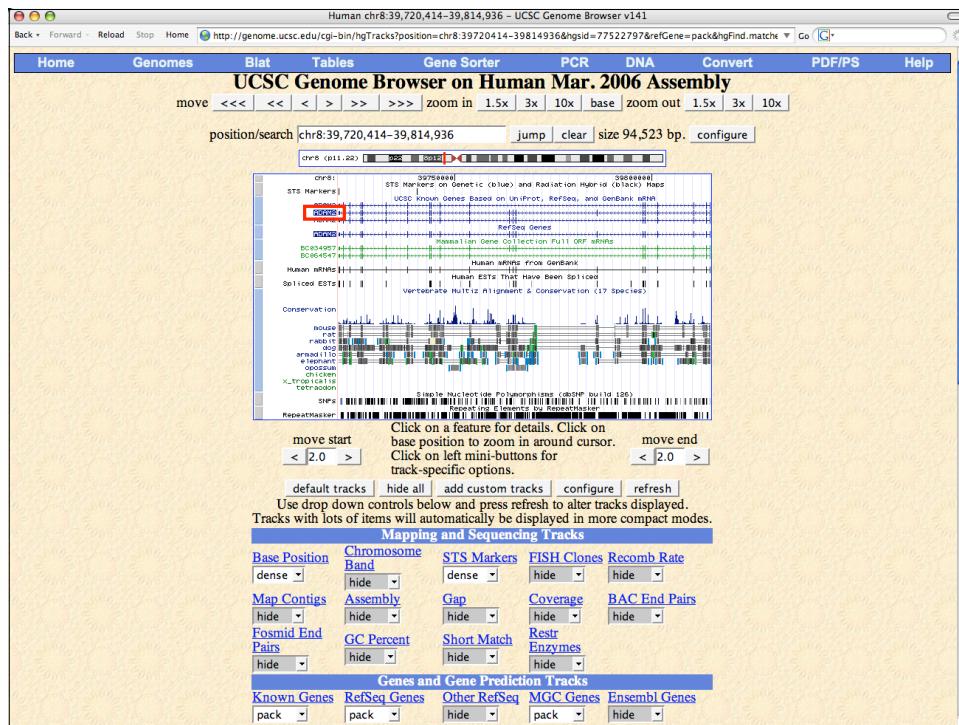
**RefSeq Genes**

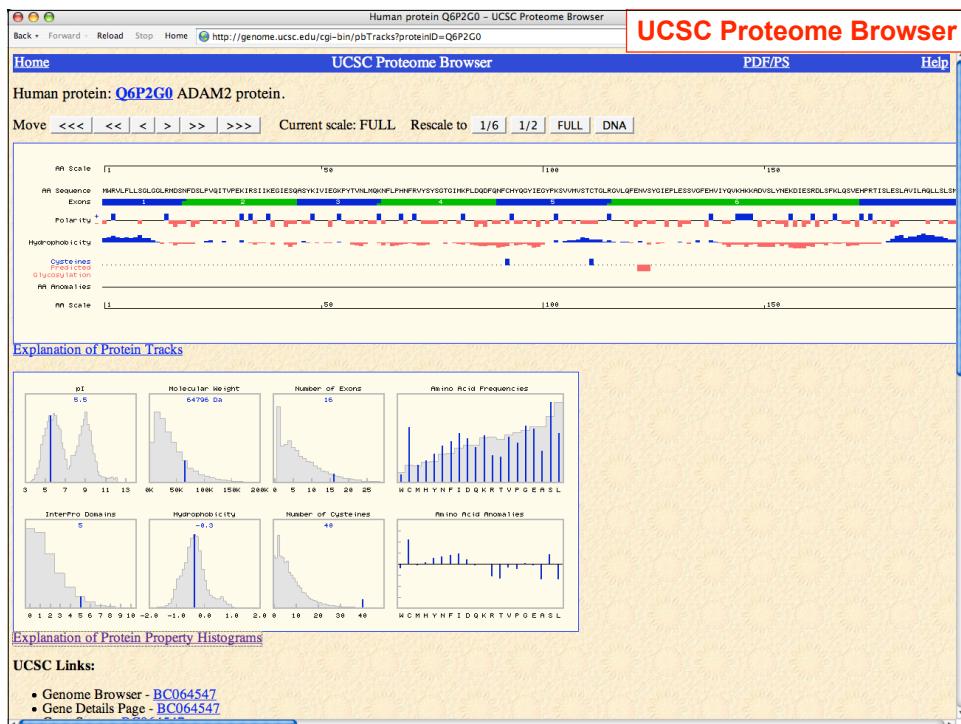
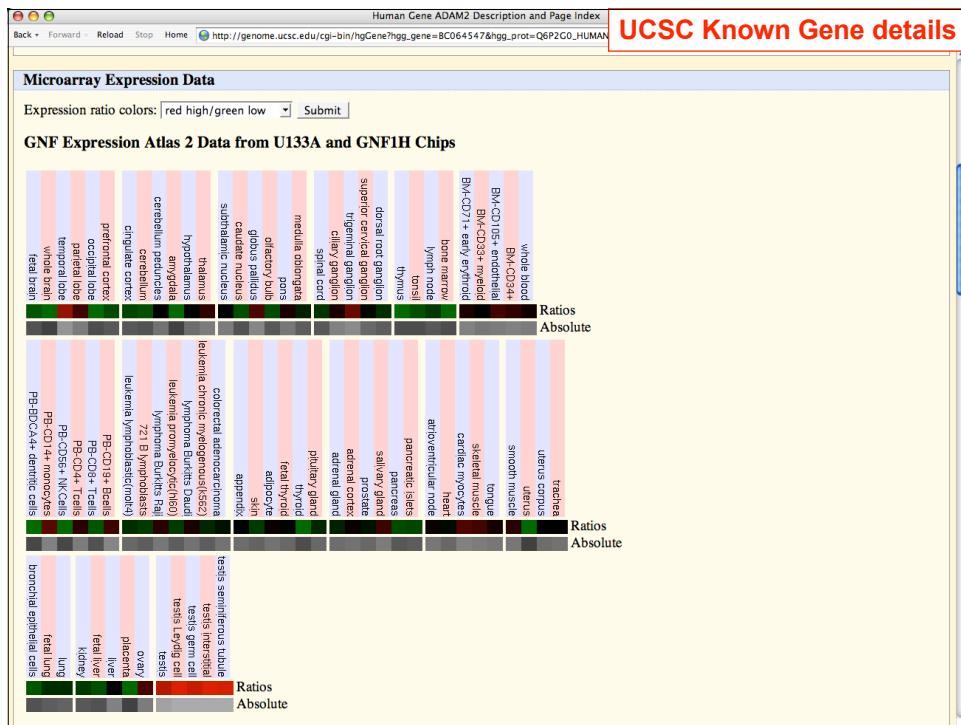
ADAM2 at chr8:39720414-39814936 - (NM\_001464) ADAM metallopeptidase domain 2 proprotein  
 ADAM20 at chr14:70058832-70071485 - (NM\_003814) ADAM metallopeptidase domain 20 preprotein  
 ADAM21 at chr14:69993970-69996374 - (NM\_003813) ADAM metallopeptidase domain 21 preprotein  
 ADAM22 at chr7:87401638-87649364 - (NM\_004194) ADAM metallopeptidase domain 22 isoform 4  
 ADAM22 at chr7:87401638-87649383 - (NM\_021722) ADAM metallopeptidase domain 22 isoform 5  
 ADAM22 at chr7:87401638-87649383 - (NM\_021722) ADAM metallopeptidase domain 22 isoform 1  
 ADAM22 at chr7:87401638-87649383 - (NM\_021722) ADAM metallopeptidase domain 22 isoform 3  
 ADAM22 at chr7:87401638-87649383 - (NM\_016351) ADAM metallopeptidase domain 22 isoform 2  
 ADAM22 at chr7:87401638-87649383 - (NM\_016351) ADAM metallopeptidase domain 23 preprotein  
 ADAM22 at chr7:87401638-87649383 - (NM\_021722) ADAM metallopeptidase domain 28 isoform 3  
 ADAM28 at chr8:24207525-24249555 - (NM\_021777) ADAM metallopeptidase domain 28 isoform 1  
 ADAM28 at chr8:24207525-24249555 - (NM\_021777) ADAM metallopeptidase domain 28 isoform 3  
 ADAM28 at chr8:24207525-24249555 - (NM\_021777) ADAM metallopeptidase domain 28 isoform 1  
 ADAM29 at chr4:176076134-176135905 - (NM\_014269) ADAM metallopeptidase domain 29 preprotein

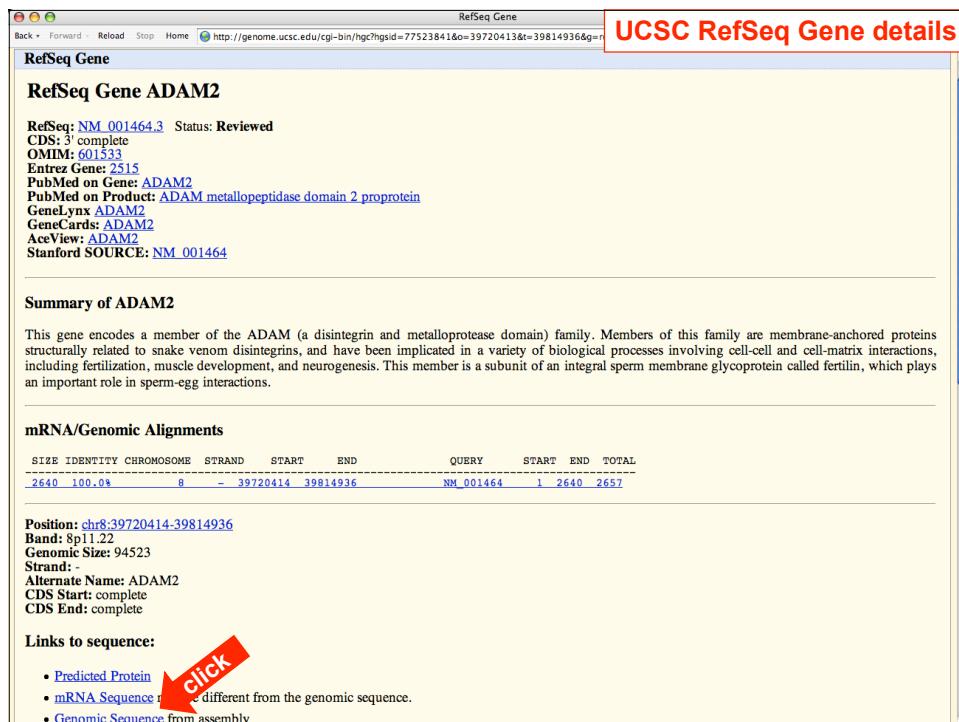
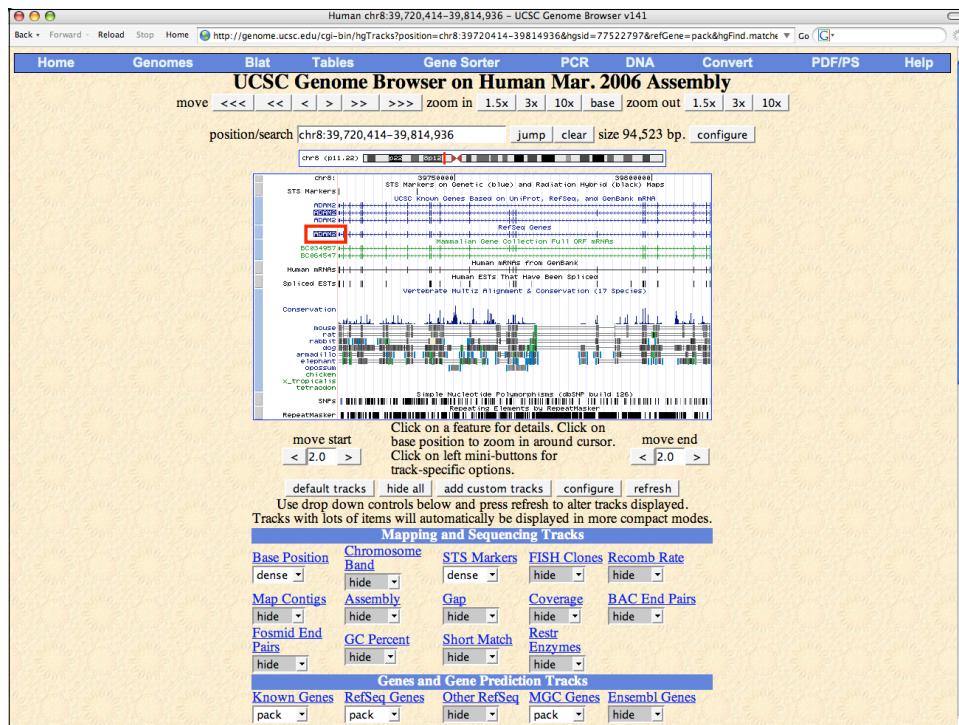
**Non-Human RefSeq Genes**

ADAM2 at Chr8:39720414-39814902 - (NM\_213967) ferteilin beta  
 ADAM2 at Chr8:39720414-39814988 - (NM\_174220) a disintegrin and metalloprotease domain 2  
 Adam2 at Chr8:39720414-39801569 - (NM\_009618) a disintegrin and metalloprotease domain 2  
 Adam2 at Chr8:39723147-39913877 - (NM\_020077) a disintegrin and metalloprotease domain 2  
 Adam21 at Chr14:699898547-69996354 - (NM\_020330) disintegrin and metalloprotease domain 21  
 Adam21 at Chr14:699898547-69996354 - (NM\_020330) disintegrin and metalloprotease domain 21  
 Adam22 at Chr7:87401638-87649364 - (NM\_004194) ADAM metallopeptidase domain 22  
 Adam22 at Chr7:87401638-87649383 - (NM\_021722) ADAM metallopeptidase domain 22  
 Adam22 at Chr7:87401638-87649383 - (NM\_021722) ADAM metallopeptidase domain 22  
 Adam23 at Chr2:207016503-207194450 - (NM\_011780) a disintegrin and metallopeptidase domain 23  
 Adam23 at Chr2:207016503-207194450 - (NM\_011780) a disintegrin and metallopeptidase domain 23  
 Adam23 predicted at Chr2:207016503-207190724 - (NM\_001029899) a disintegrin and metallopeptidase domain 23  
 Adam24 at Chr8:13956125-39706780 - (NM\_003814) a disintegrin and metallopeptidase domain 24  
 Adam24 at Chr8:13956125-39706780 - (NM\_003814) a disintegrin and metallopeptidase domain 24  
 Adam24 at Chr14:70058832-70060969 - (NM\_010086) a disintegrin and metallopeptidase domain 24  
 Adam24 at Chr14:70058832-70060969 - (NM\_010086) a disintegrin and metallopeptidase domain 24  
 Adam25 at Chr4:18890441-188906248 - (NM\_011781) a disintegrin and metallopeptidase domain 25  
 Adam25 at Chr4:18890441-188906248 - (NM\_011781) a disintegrin and metallopeptidase domain 25  
 Adam25 at Chr14:699898547-69996354 - (NM\_020330) disintegrin and metalloprotease domain 25  
 Adam25 at Chr14:699898547-69996354 - (NM\_020330) disintegrin and metalloprotease domain 25  
 Adam26a at Chr8:17371523-17373287 - (NM\_010085) a disintegrin and metallopeptidase domain 26  
 Adam26a at Chr8:17371523-17373287 - (NM\_010085) a disintegrin and metallopeptidase domain 26

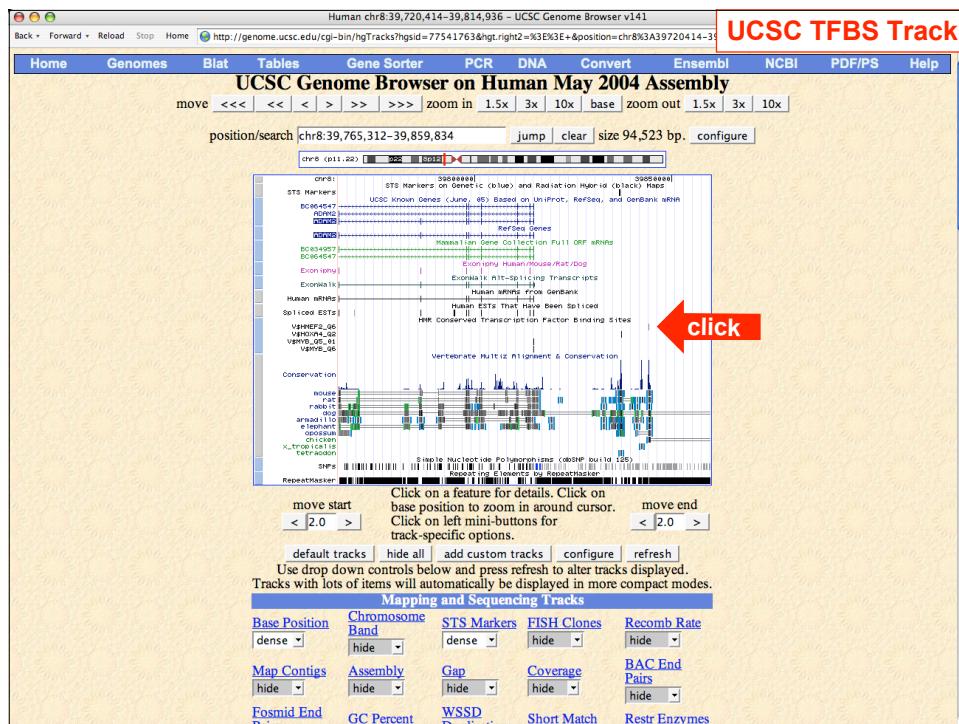
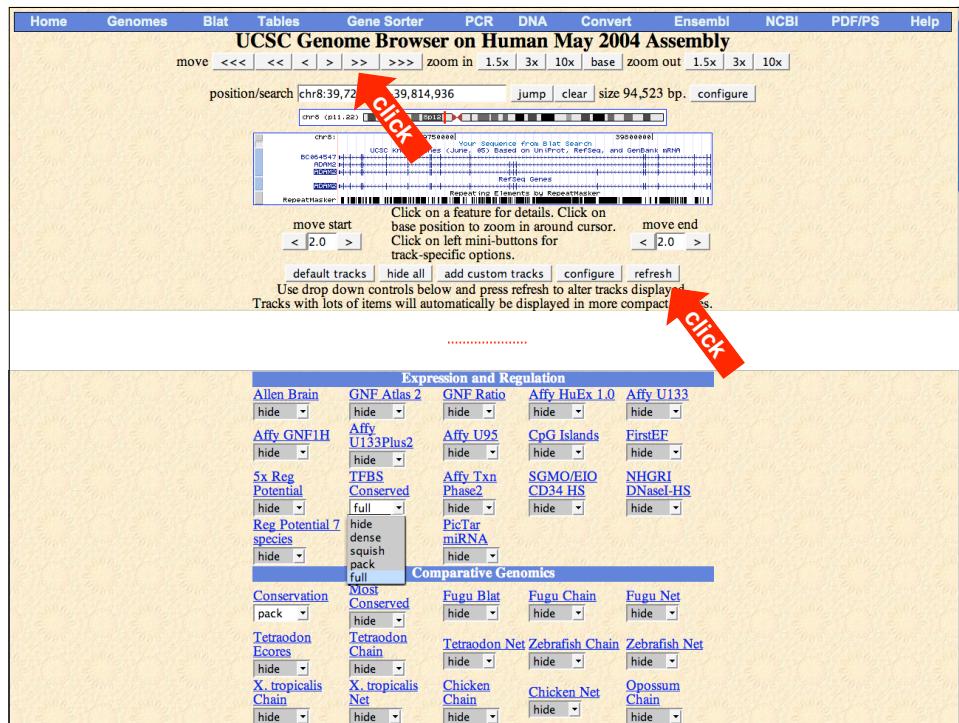
NHGRI Current Topics in Genome Analysis 2006  
 Mining Genomic Sequence Data











HMR Conserved Transcription Factor Binding Sites (V\$HMEF2\_Q6)

**UCSC TFBS Track details**

Home Genomes Genome Browser Blat Tables Gene Sorter PCR FAQ Help

HMR Conserved Transcription Factor Binding Sites (V\$HMEF2\_Q6)

**Transcription Factor Binding Site information:**

Item: V\$HMEF2\_Q6  
Transfac matrix link: [M00406](#)  
Score: 874  
Zscore: 3.50  
Strand: -  
Position: [chr8:39844166-39844181](#)  
Band: 8p11.22  
Genomic Size: 16  
[View DNA for this feature](#)

---

**Transcription Factors known to bind to this site:**

Factor: MEF-2A  
Species: human  
SwissProt ID: Q02078  
[Proteome Browser Entry](#)  
[View table schema](#)

Data last updated: 2004-12-14

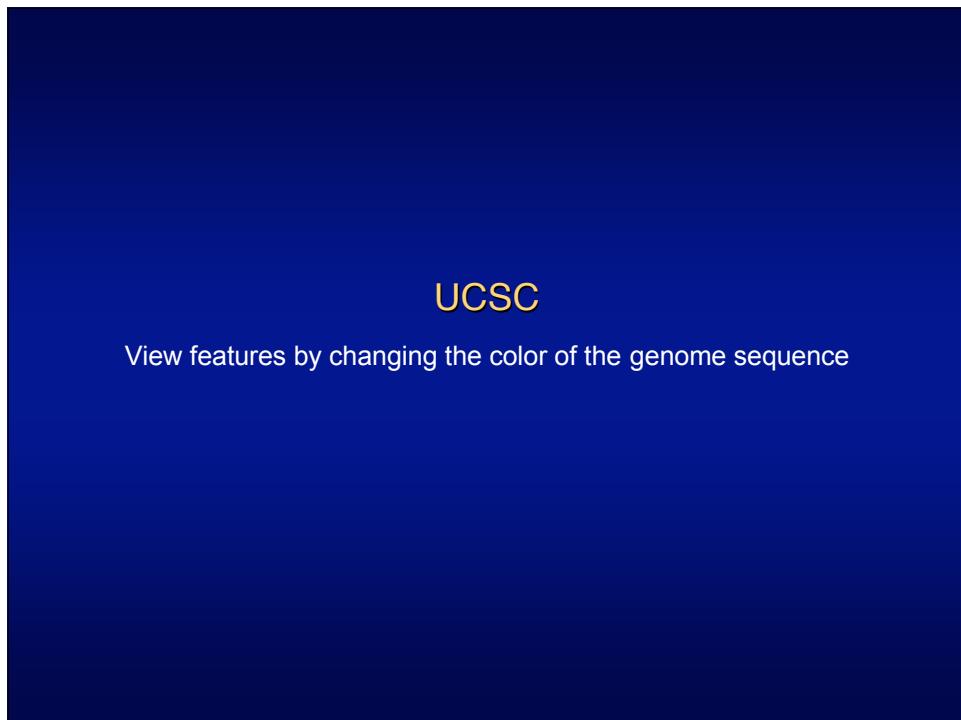
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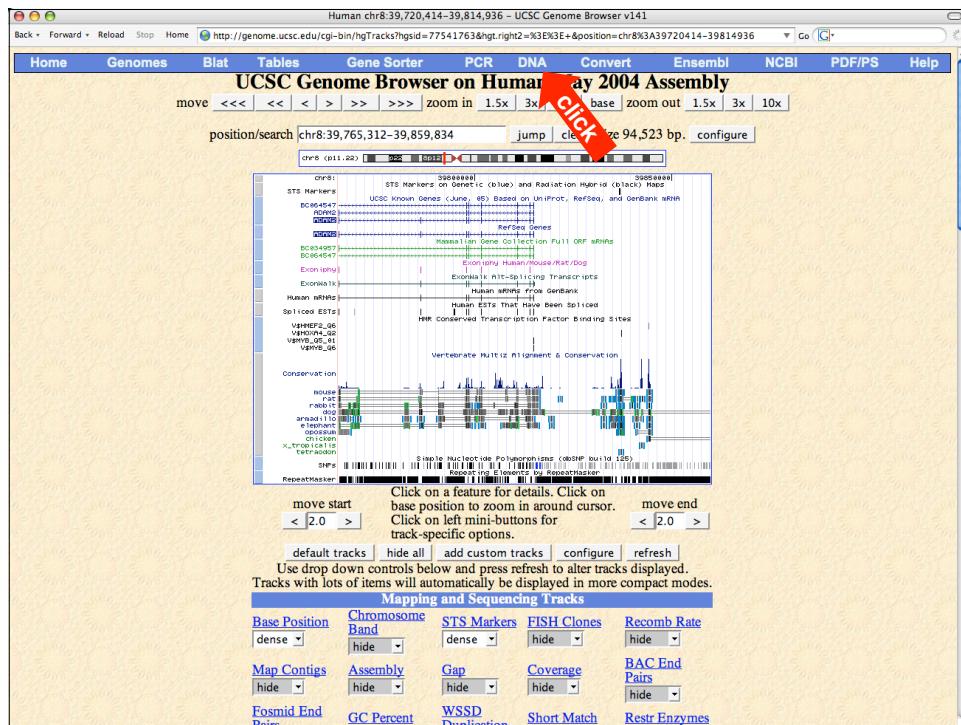
**Description**

This track contains the location and score of transcription factor binding sites conserved in the human/mouse/rat alignment. A binding site is considered to be conserved across the alignment if its score meets the threshold score for its binding matrix in all 3 species. The score and threshold are computed with the Transfac Matrix Database (v8.3) created by [Biobase](#). The data are purely computational, and as such not all binding sites listed here are biologically functional binding sites.

In the graphical display, each box represents one conserved tfbs. Clicking on a box brings up detailed information on the binding site, namely its Transfac I.D., a link to its Transfac Matrix (free registration with Transfac required), its location in the human genome (chromosome, start, end, and strand), its length in bases, its raw score, and its Z score.

All binding factors that are known to bind to the particular binding matrix of the binding site are listed along with their species, SwissProt ID, and a link to that factor's page on the UCSC Protein Browser if such an entry exists.

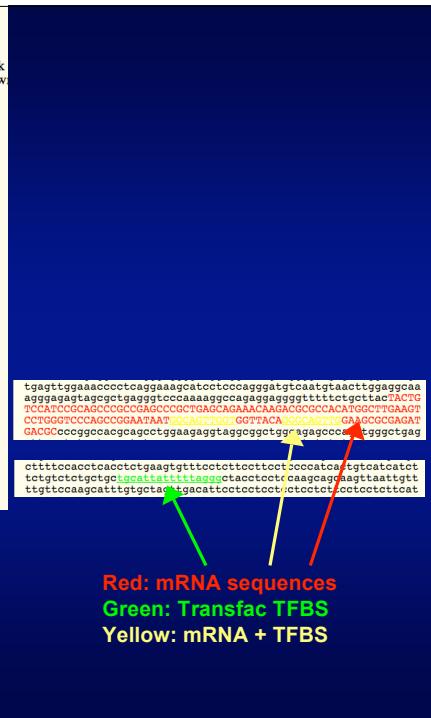


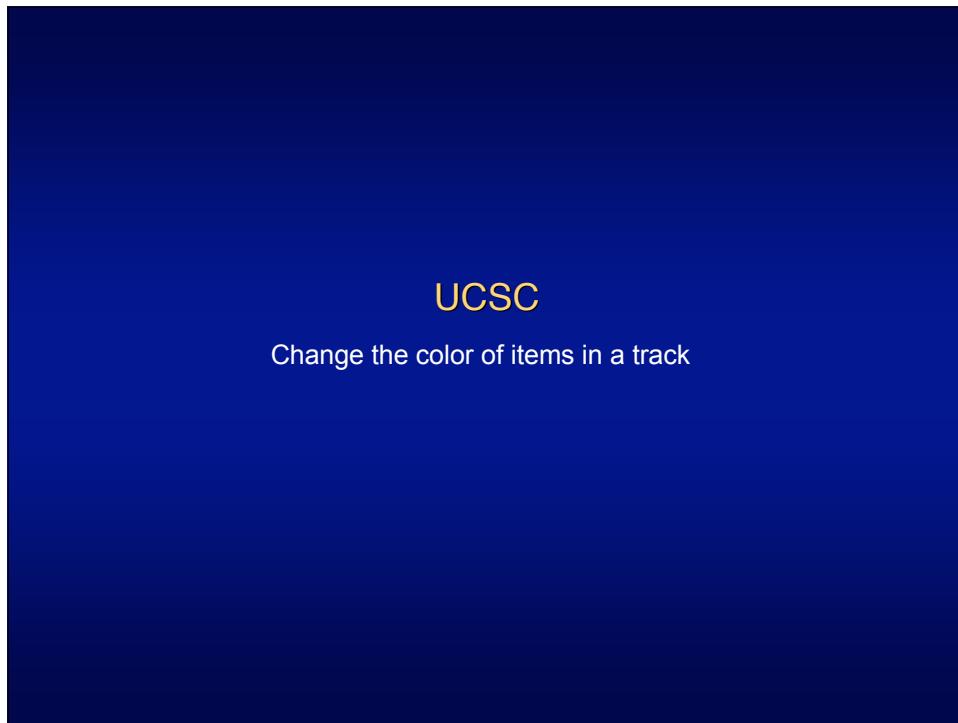


## Extended DNA Case/Color Options

Use this page to highlight features in genomic DNA text. DNA covered by a particular track below for details about color, and for examples. Tracks in "hide" display mode are not shown.

Position: chr8:39,765,312-39,859,834	Reverse complement <input type="checkbox"/>						
Letters per line: 60	Default case: <input checked="" type="radio"/> Upper <input type="radio"/> Lower						
<input type="button" value="submit"/>							
Track Name	Toggle Underline	Bold	Italic	Red	Green	Blue	
Chromosome Band (Ideogram)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
STS Markers	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Known Genes	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	255	0	0
RefSeq Genes	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	255	0	0
MGC Genes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	0	0	0
Vega Genes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	0	0	0
Vega Pseudogenes	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	0	0	0
Exoniphy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	0	0	0
Human mRNAs	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	255	0	0
Spliced ESTs	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	0	0	0
ExonWalk	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	0	0	0
TFBS Conserved	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	0	255	0
Conservation	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	0	0	0
SNPs	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	0	0	0
RepeatMasker	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	0	0	0





Human chr8:39,720,414–39,814,936 – UCSC Genome Browser v142

Back • Forward • Reload Stop Home http://genome.ucsc.edu/cgi-bin/hgTracks Go

position/search chr8:39,720,414–39,814,936 jump clear size 94,523 bp. configure

chr8 (p11.2) 39,814,936 39,720,414

UCSC Known Genes Based on UniProt, RefSeq, and Gencode genes

RefSeq Genes

Human mRNAs Human ESTs That Have Been Spliced

Spliced ESTs Human mRNAs From Genbank

Conservation Vertebrate Multiple Alignment & Conservation (17 species)

SNPs Single Nucleotide Polymorphisms (dbSNP build 128)

RepeatMasker Click on a feature for details. Click on base position to zoom in around cursor.

move start move end

< [2.0] > < [2.0] >

Click on left mini-buttons for track-specific options.

default tracks hide all add custom tracks configure refresh

Use drop down controls below and press refresh to alter tracks displayed.

Tracks with lots of items will automatically be displayed in more compact modes.

Mapping and Sequencing Tracks

Base Position	Chromosome	STS Markers	FISH Clones	Recomb Rate
dense	Band	hide	hide	hide
Map Contigs	Assembly	Gap	Coverage	BAC End Pairs
hide	hide	hide	hide	hide

.....

Variation and Repeats

SNPs	Segmental Dups	RepeatMasker	Simple	Microsatellite
dense	hide	dense	hide	hide
Self Chain	hide			

refresh

A red arrow points to the 'SNPs' button in the Variation and Repeats section.

**UCSC SNP Track details**

**SNPs Track Settings**

## Simple Nucleotide Polymorphisms (dbSNP build 126)

Display mode: Full

Minimum Average Heterozygosity: 0  
 Maximum Weight: 3

Any type of data can be excluded from view by deselecting the checkbox below. Not all assemblies include values in all categories.

**Location Type:**  
 Unknown  Range  Exact  Between  RangeInsertion  RangeSubstitution  RangeDeletion

**Class:**  
 Unknown  Single Nucleotide Polymorphism  In/Del  Heterozygous  Microsatellite  Named  No Variation  Mixed  Mnp  Insertion  Deletion

**Validation:**  
 Unknown  By Cluster  By Frequency  By Submitter  By 2 Hit / 2 Allele  By HapMap

**Function:**  
 Unknown  Locus  Coding - Synonymous  Coding - Non-Synonymous  Untranslated  Intron  Splice Site  Reference (coding)

**Molecule Type:**  
 Unknown  Genomic  cDNA

**SNP Feature for Color Specification:** Function

The selected feature above has the following values below. For each value, a selection of colors is available.

**Unknown:** black  **Locus:** black  **Coding - Synonymous:** green  **Coding - Non-Synonymous:** red  **Untranslated:** black  **Intron:** black   
**Splice Site:** black  **Reference (coding):** black

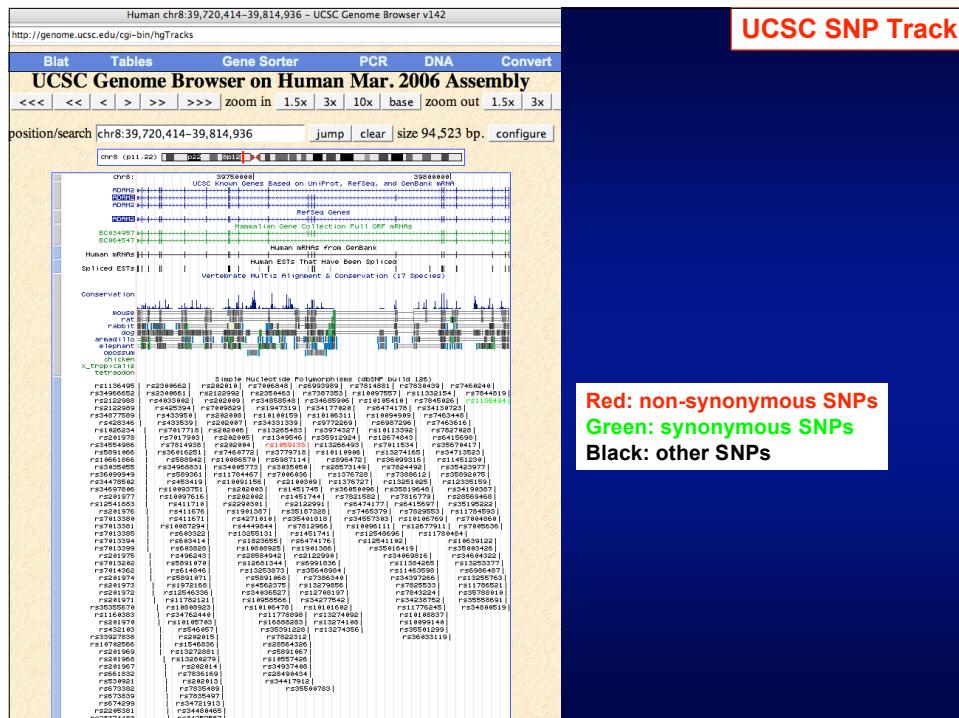
[View table schema](#)

---

**Description**

This track contains dbSNP build 126, available from [ftp.ncbi.nih.gov/snp](http://ftp.ncbi.nih.gov/snp).

**Interpreting and Configuring the Graphical Display**





**NCBI Entrez Protein**

Search | Protein  Go | Clear | Save Search

Display: **Fasta** Show: 20 Sort by Relevance Send to:

All: 1 Summary ASN.1 Related Structures: 1

1: NP\_001455 Reports ADAM metallopeptidase domain 2 proprotein [Homo sapiens] [gi:55743080]

Range: from **begin** to **end** Refresh

```
>gi|55743080|ref|NP_001455.3| ADAM metallopeptidase domain 2 proprotein [Homo sapiens]
MWRVLFILSLGCGLRMSDNFDSLPLWQI7VPKEKIRSIIKEQIBSQASVYKIVIECKPPTVNLQKQFLPLPNF
RVVSYSTGQIMKPLDQDFQNTCHYQCYILEGYPKSVWVNSTCTCLGLGVGLQFENVSYCIEPLESSVGFBVVI
RQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQV
L7NA1FVSNFTIITLSELENDENKIATTGEANELLHTFLRNWKTSTYLV1LAPHDVAFLVVREKSNVYGA
TFOCKMKCDANYAGCVCVLIJHPRTISLESLAVLAQQLSLEMGIC1YD1NKCQCQAGAVCINNPEA1HFSGVK1
PESQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQVQV
FKAGSNCA2BGPCCENC1LMSKERMCRPSFEECDLREYCNSSASCPEHHVYQTOUPCGLNQWIC1DGVMC
SGDKQCCTDFGKEVBPGECKSYHLNSKTDVSGNC1C1SDGQYTCQADANLQCGKLICKYVGKFLLQJIPRA
T1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1L1
K1C1C1C1S1YLPDCDSVQ8DLNPGCS1D5GNPPPVIA1A1P1P1P1V1D1V1H1H1P1H1P1H1P1H1P1H1P1
V1L1A1M1V1K1N1F1Q1R1K1W1T1E1D1Y1S1D1B1Q1P1E1P1S1E1C1G1O1C1T1R1C1N1G1V1N1P1
```

**Chicken BLAT Search**

**UCSC BLAT search**

Home Genomes Tables Gene Sorter FAQ Help

**Chicken BLAT Search**

**BLAT Search Genome**

Genome: Chicken Assembly: Feb. 2004 Query type: BLAT's guess Sort output: query,score Output type: hyperlink

```
>gi|55743080|ref|NP_001455.3| ADAM metallopeptidase domain 2 proprotein [Homo sapiens]
MQLGGLRHSNPFSLPVQITPKEKRSIIKKGIESQASYKIVIBGRKYTVLMLQONFLPHNF
RVVYSGCTGIMKMPPLDDDFPNFCHVCGVIBGVPSVWVSTCIGLRGVLFQENVSYGIEPLSSSVCFBHV
YQVKKHRADVSLYNKEDIDESRDLSPFKLQSVPEODFAYKIEHHVITVEKLQYHMSDPTVVAKVFPOLIG
L/TNAIPVSFNNTITILLSLLEINDEKLAATGEANELLFTFLWKWTSYLVLRPHDVAFLLVYREKSNYVGA
TFOGKMKCDANYAGGVVHLHETVLSLESLAVIAQLLSEMGITYDANKCCSGATVMMPEAHLHSGVKI
PFGKQKQGKQKQGKQKQGKQKQGKQKQGKQKQGKQKQGKQKQGKQKQGKQKQGKQKQGKQKQGKQKQGKQKQGKQ
PKXAGSMCACBGPCCENCLPMSKEBMRMCRSFPECDLPYEYCNSSASCPENHVQVGHPCGLNQWICIDGVCVM
SGDXQQTDTFGKEVEFPGPSECYSHLNKSKTDVDSGNCGIDSQGYTCCEADNLQCGKLICKYVGKFLLQJPR
TIIYANISGHLICIAVEFASDHADSGRMWIKDGTSGSNKVCRNRCRVSSSYLYGVDCTTDKNDRGCVNKK
KCHCISASYLPPDCSVQSDLNPQGSIDSGNFPVPAIPARLPERRYIENIYHSKPMRWPFFLPIFFFIIFC
VLIAINVRVNQQRKWRTEDSSDQEPESEEPKG
```

Paste in a query sequence to find its location in the genome. Multiple sequences may be searched if separated by lines starting with > followed by the sequence name.

**File Upload:** Rather than pasting a sequence, you can choose to upload a text file containing the sequence.  
 Upload sequence:

Only DNA sequences of 25,000 or fewer bases and protein or translated sequence of 10000 or fewer letters will be processed. Up to 25 sequences can be submitted at the same time. The total limit for multiple sequence submissions is 50,000 bases or 25,000 letters.

For locating PCR primers, use [In-Silico PCR](#) for best results instead of BLAT.

**About BLAT**

BLAT on DNA is designed to quickly find sequences of 95% and greater similarity of length 40 bases or more. It may miss more divergent or shorter sequence alignments. It will find perfect sequence matches of 33 bases, and sometimes find them down to 20 bases. BLAT on proteins finds sequences of 80% and greater similarity of length 20 amino acids or more. In practice DNA BLAT works well on primates, and protein BLAT on land vertebrates.

**Chicken BLAT Results**

**UCSC BLAT search**

Home Genomes Tables Gene Sorter PCR FAQ Help

**Chicken BLAT Results**

**BLAT Search Results**

ACTIONS	QUERY	SCORE	START	END	QSIZE	IDENTITY	CHRO	STRAND	START	END	SPAN	
<a href="#">broswer</a>	<a href="#">details</a>	NP_001455..3	44	539	600	735	71.6%	0n	++	635370	635555	186
<a href="#">broswer</a>	<a href="#">details</a>	NP_001455..3	12	301	304	735	100.0%	0	++	67659709	67659720	12
<a href="#">broswer</a>	<a href="#">details</a>	NP_001455..3	12	437	440	735	100.0%	1	++	67660117	67660128	12
<a href="#">broswer</a>	<a href="#">details</a>	NP_001455..3	12	385	390	735	83.4%	1	++	67659961	67659978	18

**UCSC Genome Browser on Chicken Feb. 2004 Assembly**

move <<< << < > >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x

position/search [chrUn:635,370-635,555](#) jump clear size 186 bp. configure

Click on a feature for details. Click on base position to zoom in around cursor. move end  
 Click on left mini-buttons for track-specific options.

**Chromosome Color Key:** 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9 | 10 | 11 | 12 | 13 | 14 | 15 | 16 | 17 | 18 | 19 | 20 | 21 | 22 | X | Y | M | U |  
 Use drop down controls below and press refresh to alter tracks displayed.  
 Tracks with lots of items will automatically be displayed in more compact modes.

**Mapping and Sequencing Tracks**

Base Position	Supercontigs	Assembly	Gap	BAC End Pairs
dense	hide	hide	hide	hide
GC Percent	Quality Scores	Isochores	Contamination	Short Match
hide	hide	hide	hide	hide

UCSC BLAT search

ACTIONS	QUERY	SCORE	START	END	QSIZE	IDENTITY	CHRO	STRAND	START	END	SPAN
browser details	NP_001455.3	44	539	600	735	71.6%	Un	++	635370	635555	186
browser details	NP_001455.3	12	301	304	735	100.0%	1	++	67659709	67659720	12
browser details	NP_001455.3	12	437	440	735	100.0%	1	++	67660117	67660128	12
browser details	NP_001455.3										

**Alignment of NP\_001455.3 and chrUn:635370-635555**

Click on links in the frame to the left to navigate through the alignment. Matching bases are colored blue and sequence.

**NP\_001455.3**

NP\_001455.3  
 Chicken.chrUn  
 block1  
 block2  
 block3  
 together

```

mrvrliflq lgeqlmdnenf delpvgitvp okirsiikieg iegsgasykv iiegkpytvnl 60
mgknflphnf rrvsysstgq mhpqdqdfn fchyggyies ypksvvwmq ctglgrvlgf 120
dgvqsaqtdt qmnmavlmw uqiwmsvmt sasqfgrkqntv qygttqgkqf 180
emhivivqpl yhnngsdktv vqgkvtqflig ltnaihwfsm fttileqel wldenklat 240
geanolllhtl iwktsayvl rphdvalflv yrskenvyga tfiqkmnodan yagggvhlpr 300
tialalavt lqdlalang lystcdqg gavavvqg odqnhsgqg fmgnlghd 360
mlakgeqoc rkcspcecp tltawcpcp klesweede atetccgqgrqf accidater 420
fkagsndae pecencilmfa kermcrpafe ecclipeyeng aasacpenhy vqtghepgln 480
qcdtideqv spqhdccitf gkevngpse cyshnsihv viagnyciida gyqgeqedL 540
qcGEKccavv gqdcnttq uqgvscuqf vqgkqf vqgkqf qf gqgkqf qf gqgkqf 600
erngrcvwss ylygdtttdk endrygvnck khchssasyi ppdcswqds1 wpqgsidapn 660
fpvpaipar perriyienly hekpmrwplf llifipffile vliainvkx 720
yeddeqgees qeqhg
    
```

**Chicken.chrUn :**

```

AACTGCGGT GGGGAAACT CATCCTCaca TA>ccaaatc gapttcctt caccaattta 635429
aatgt ACCA TCATCATGc Tcaatgtgc aaacATCTGT G>qgttcctt tttatgtatataat 635469
catgacccctt caggggccata ttccatctcg gttAGGGATA GCACGAAATG CCGTcccggaa 635549
AAAGOTA
    
```

**Side by Side Alignment\***

```

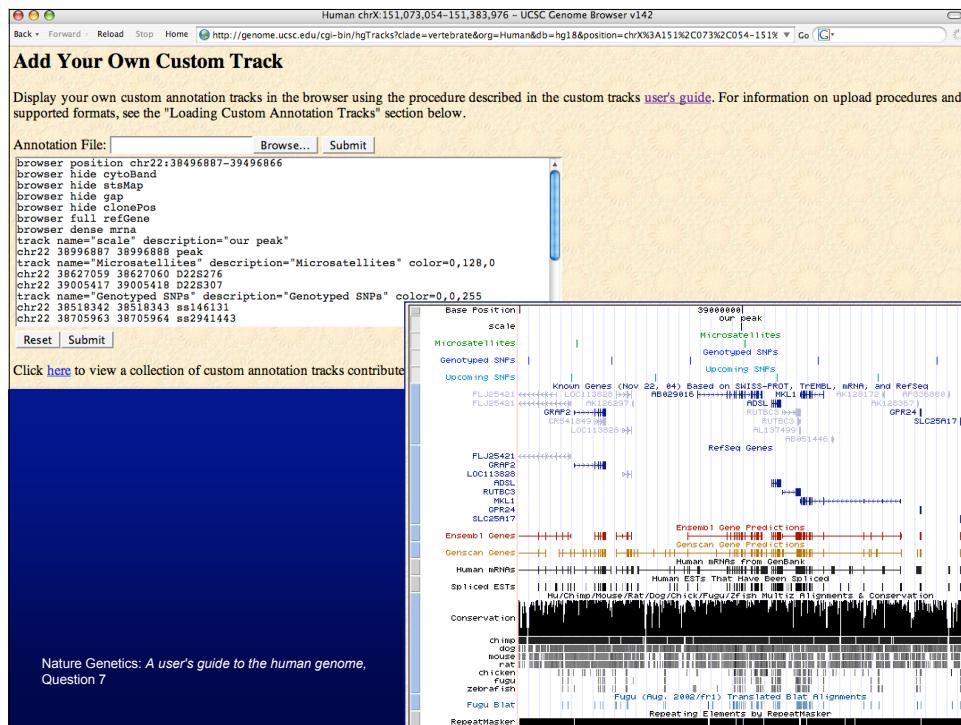
001615 N L Q C C K L I Q K Y 001647
>>>> 635370 aactcggggotgtggaaaactcatgtcacataac 635402

001681 T I I Y A N I S G H L C 001716
>>>> 635436 accatcaactatgtcaagtgcaagacatcgtgtc 635471

001768 X D G T S C G S N K V 001800
>>>> 635523 aaggatggcacgaaatgccccgttccggaaaggta 635555
    
```

UCSC

Add your own custom tracks



## UCSC Table Browser

- Download track in text format
- Retrieve DNA sequence covered by a track
- Calculate intersections between tracks and view in the Genome Browser. For example:
  - Show all RefSeq genes that contain only one exon
  - Show transcription factor binding sites that overlap (intersect) with a SNP

**Table Browser**

Use this program to get the data associated with a track in text format, to calculate intersections. See [Using the Table Browser](#) for a description of the controls in this form.

clade: Vertebrate genome: Human assembly: Mar. 2006

group: Genes and Gene Prediction Tracks track: RefSeq Genes

table: refGene describe table schema

region: genome position chr22:38496887-39496866 lookup

identifiers (names/acceessions): paste list upload list

filter:

intersection:

correlation:

output format: all fields from selected table

output file: (leave blank to keep output)

file type returned: plain text gzip compressed

[get output](#) [summary/statistics](#)

To reset all user cart settings (including custom tracks), [click here](#).

**UCSC Table Browser: RefSeq genes that contain only one exon**

**Filter on Fields from hg18.refGene**

bin	is ignored	match	AND
name	does	match	AND
chrom	does	match	AND
strand	does	match	AND
txStart	is ignored	match	AND
txEnd	is ignored	match	AND
cdsStart	is ignored	match	AND
cdsEnd	is ignored	match	AND
exonCount	is =	1	AND
exonStarts	does	match	
exonEnds	does	match	
id	is ignored	match	AND
name2	does	match	AND
cdsStartStat	does	match	AND
cdsEndStat	does	match	AND
exonFrames	does	match	

AND - Free-form query:

[submit](#) [cancel](#)

A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P		
1	#filter: refGene.exonCount = 1																
2	#bin	name	chrom	strand	txStart	txEnd	cdsStart	cdsEnd	exonCount	exonStarts	exonEnds	id	name2	cdsStartStat	cdsEndStat	exonFrames	
3	NM_0006511	chr1	-	15858910	15864803	15859593	15860893	1	15859593	15860893	0	R5C1A1		incmpl	0		
4	178	NM_002232	chr1	-	111015932	111019178	111017226	111018954	1	111015932	111019178	0	KCN43		cmpl	0	
5	301	NM_001821	chr1	-	239858790	239865875	239863720	239865691	1	239858790	239865855	0	CHML		cmpl	0	
6	NM_001005221	chr1	+	357521	358458	357521	358458	1	357521	358458	0	OR4F25		cmpl	0		
7	587	NM_001005221	chr1	+	357521	358458	357521	358458	1	357521	358458	0	OR4F29		incmpl	0	
8	587	NM_001005224	chr1	+	357521	358458	357521	358458	1	357521	358458	0	OR4F29		cmpl	incmpl	0
9	NM_001005224	chr1	+	357521	358458	357521	358458	1	357521	358458	0	OR4F29		cmpl	incmpl	0	
10	589	NM_001005221	chr1	-	610960	611897	610960	611897	1	610960	611897	0	OR4F29		incmpl	cmpl	0
11	589	NM_001005224	chr1	-	610960	611897	610960	611897	1	610960	611897	0	OR4F3		incmpl	cmpl	0
12	589	NM_001005227	chr1	-	610960	611897	610960	611897	1	610960	611897	0	OR4F16		incmpl	cmpl	0
13	NM_0008043	chr1	+	1157507	11575081	1157521	1158511	1	1157507	1158511	0	BIGAT6		cmpl	0		
14	607	NM_0008044	chr1	-	2927905	2928325	2928110	2929244	1	2927905	2928325	0	ACTBLX		cmpl	0	

**NCBI**

View a genomic region between two STS markers

# NHGRI Current Topics in Genome Analysis 2006

## Mining Genomic Sequence Data

NCBI Home Page

[PubMed](#) [All Databases](#) [BLAST](#) [OMIM](#) [Books](#) [TaxBrowser](#) [Structure](#)

Search All Databases  for

**SITE MAP**  
Alphabetical List  
Resource Guide

**About NCBI**  
An introduction to NCBI

**GenBank**  
Sequence submission support and software

**Literature databases**  
PubMed, OMIM, Books, and PubMed Central

**Molecular databases**  
Sequences, structures, and taxonomy

**Genomic blotters**  
the human genome, whole genomes, and related resources

**Tools**  
Data mining

**Research at NCBI**  
People, projects, and seminars

**Software engineering**  
Tools, RAD, and databases

**Education**  
Teaching resources and on-line tutorials

**FTP site**

**National Center for Biotechnology Information**  
National Library of Medicine National Institute of Health

**What does NCBI do?**

Established in 1988 as a national resource for molecular biology information, NCBI creates public databases, conducts research in computational biology, develops software tools for analyzing genome data, and disseminates biomedical information - all for the better understanding of molecular processes affecting human health and disease. [More...](#)

**Whole Genome Association**

The NCBI Whole Genome Association (WGA) resource provides researchers with access to genotype and associated phenotype information that will help elucidate the link between genes and disease. For more information, click here to see the the WGA resource page and click here to read the [press release](#).

**100 Gigabases**

GenBank and its collaborating databases, the European Molecular Biology Laboratory and the DNA Data Bank of Japan, have reached a milestone of 100 billion bases from over 165,000 organisms. See the press release or find more information on [GenBank](#).

**PubMed Central**

An archive of life sciences journals

- Free fulltext
- Over 500,000 articles from over 200 journals
- Linked to PubMed and fully searchable

Use of PubMed Central requires no registration or fee. Access it from any computer with an Internet connection.

**NCBI News**

Summer 2006 News available online

**Hot Spots**

- ▶ Assembly Archive
- ▶ Clusters of orthologous groups
- ▶ Coffee Break, Genes & Disease, NCBI Handbook
- ▶ Electronic PCR
- ▶ Entrez Home
- ▶ Entrez Tools
- ▶ Gene expression omnibus (GEO)
- ▶ Human genome resources
- ▶ Influenza Virus Resource
- ▶ Map Viewer
- ▶ dbSNP
- ▶ Mouse genome resources
- ▶ My NCBI
- ▶ ORF finder
- ▶ Rat genome resources
- ▶ Reference sequence project
- ▶ SAGEmap
- ▶ SKY/CGH database
- ▶ dbSNP

Map Viewer

[Back](#) [Forward](#) [Reload](#) [Stop](#) [Home](#) <http://www.ncbi.nlm.nih.gov/mapview/> [Go](#)

Search  for

Click on the organism name to go to the genome view

**Vertebrates**

**Mammals**

- [Bos taurus \(cow\)](#)
- [Canis familiaris \(dog\)](#)
- [Felis catus \(cat\)](#)
- [Homo sapiens \(human\) Build 36](#)
- [Macaca mulatta \(rhesus macaque\)](#)
- [Mus musculus \(mouse\) Build 36](#)
- [Mus musculus \(mouse\) Build 35](#)
- [Ovis aries \(sheep\)](#)
- [Pan troglodytes \(chimpanzee\)](#)
- [Rattus norvegicus \(rat\)](#)
- [Sus scrofa \(pig\)](#)

**Other Vertebrates**

- [Danio rerio \(zebrafish\)](#)
- [Gallus gallus \(chicken\)](#)

**Invertebrates**

**Insect**

- [Anopheles gambiae \(mosquito\)](#)
- [Apis mellifera \(honey bee\) Amel\\_4.0](#)
- [Apis mellifera \(honey bee\) Amel\\_2.0](#)
- [Drosophila melanogaster \(fruit fly\)](#)
- [Tribolium castaneum \(red flour beetle\)](#)

**Nematode** [BLAST](#)

*Ceutorhynchus purpuratus* (nematode)

**Echinoderms** [BLAST](#)

*Strongylocentrotus purpuratus* (purple sea urchin)

**Protozoa** [BLAST](#)

- [Cryptosporidium parvum](#)
- [Dictyostelium discoidium](#)
- [Plasmodium falciparum](#)

See more about [Bacteria](#) [BLAST](#), [Organelles](#) [BLAST](#), [Viruses](#) [BLAST](#)

The Map Viewer supports search and display of genomic information by chromosomal position. Regions of interest can be retrieved by text queries (e.g. gene or marker name) or by sequence alignment (BLAST). View results at the whole genome level, and select what to display in more detail. Multiple options exist to configure your display, download data, navigate to related data, and analyze supporting information using the tools provided. [More...](#)

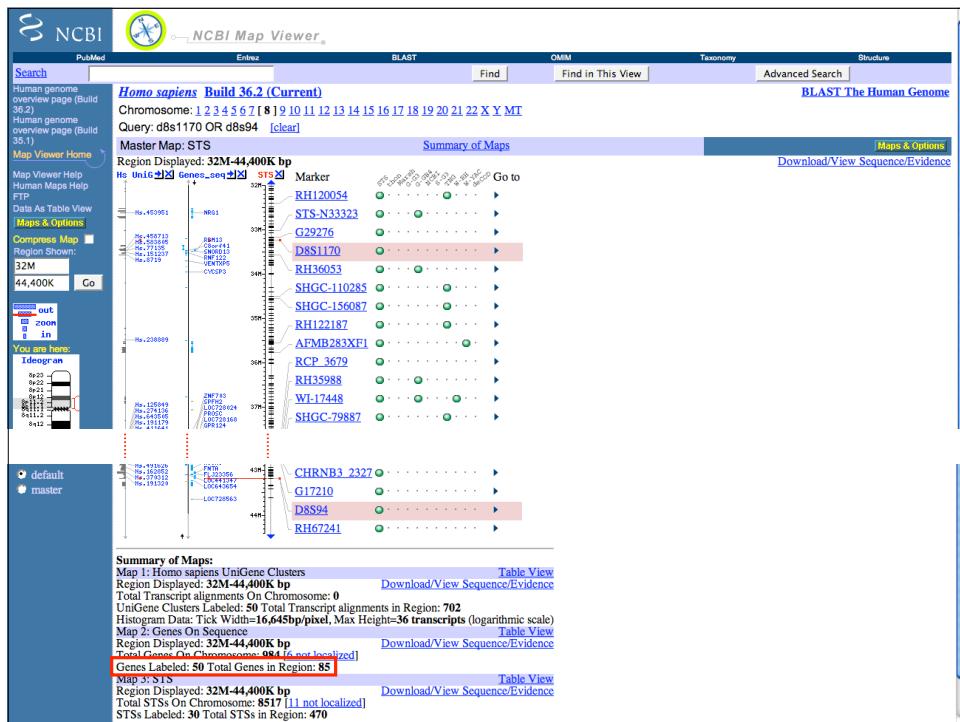
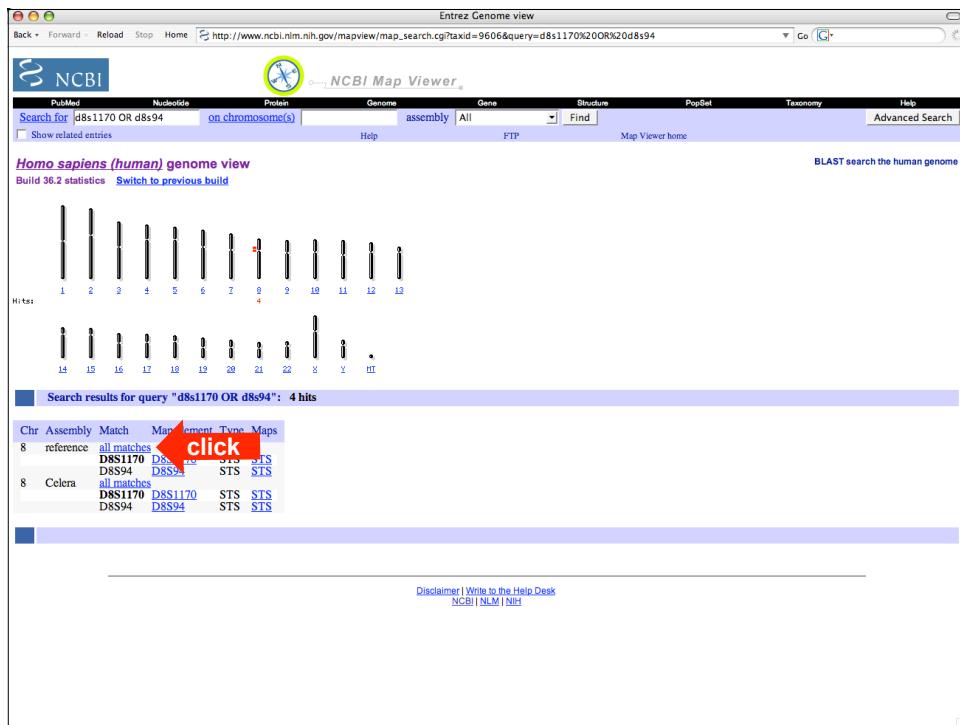
**Plants** [BLAST](#) [Search all plant maps](#)

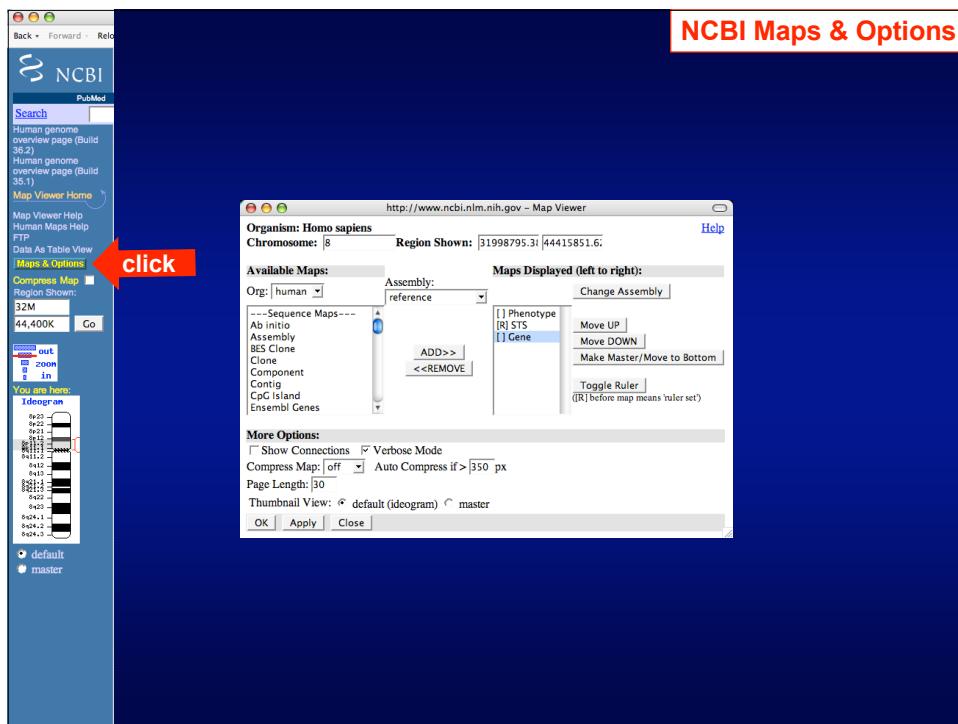
- [Arabidopsis thaliana \(thale cress\)](#)
- [Avena sativa \(oat\)](#)
- [Beta vulgaris \(beet\)](#)
- [Cicer arietinum \(soybean\)](#)
- [Hordeum vulgare \(barley\)](#)
- [Lotus japonicus \(lotus\)](#)
- [Lycopersicon esculentum \(tomato\)](#)
- [Manihot esculenta \(cassava\)](#)
- [Oryza sativa \(rice\)](#)
- [Triticum aestivum \(wheat\)](#)
- [Zea mays \(corn\)](#)

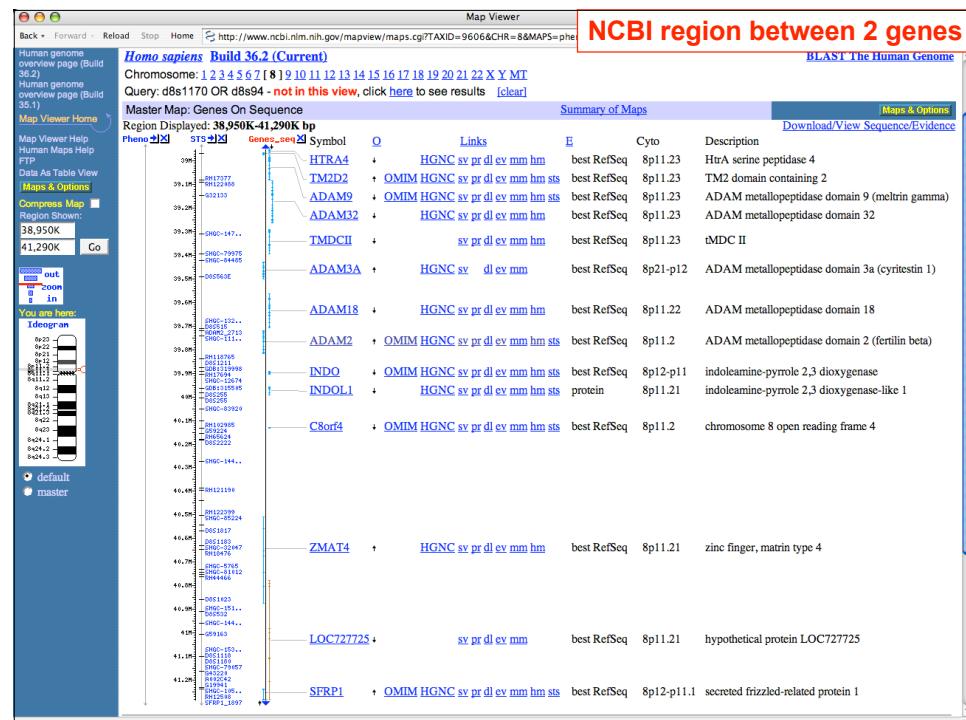
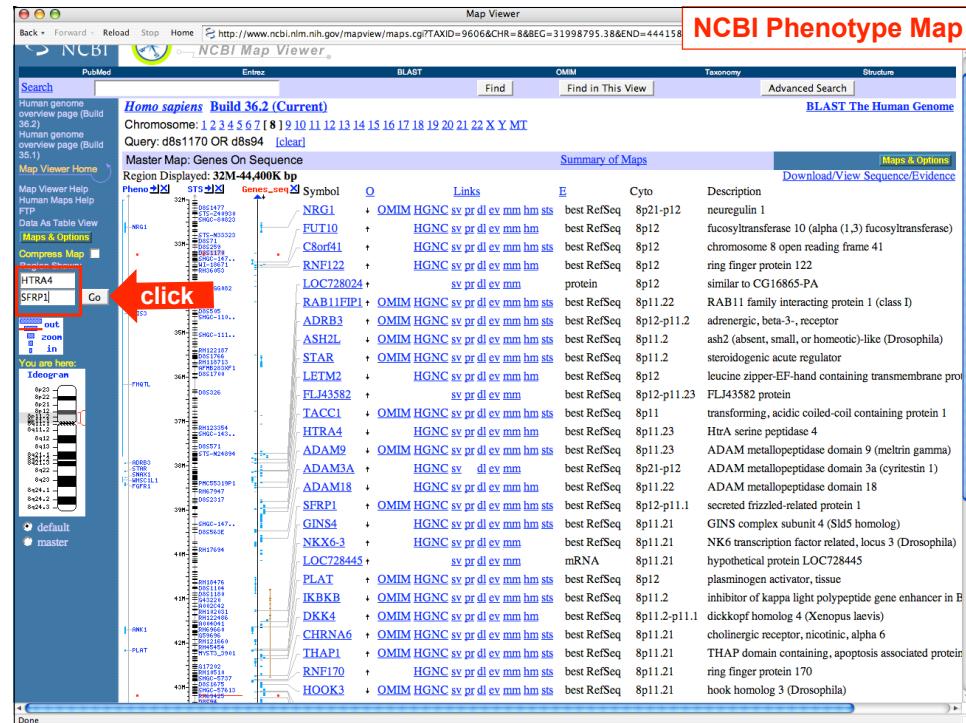
**Fungi** [BLAST](#) [Search all fungi maps](#)

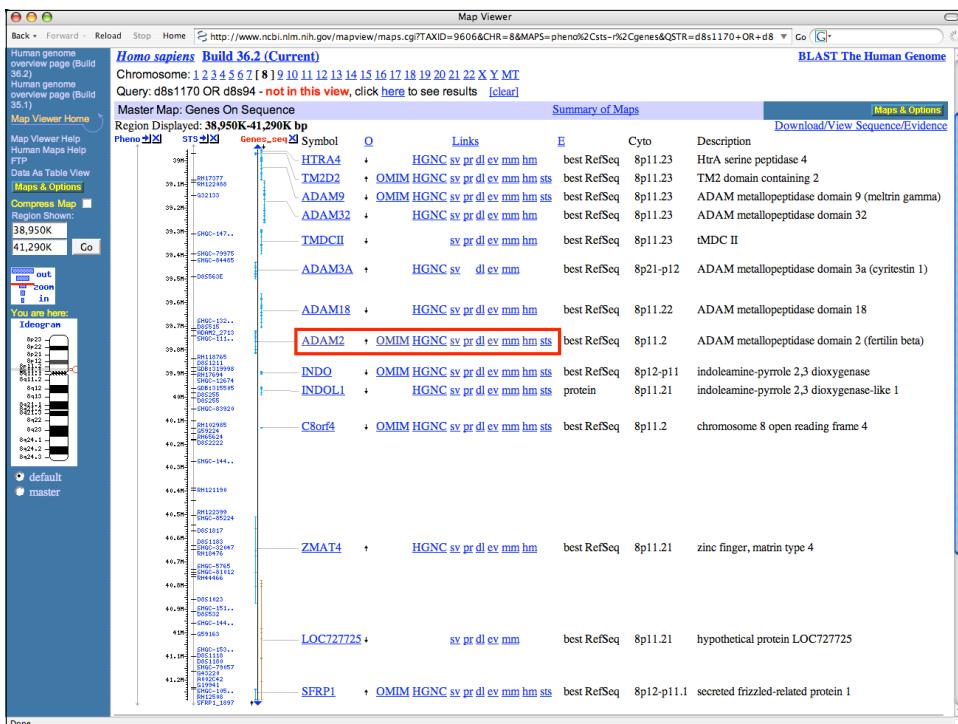
- [Aspergillus fumigatus](#)
- [Candida glabrata](#)
- [Cryptococcus neoformans](#)
- [Debaryomyces hansenii](#)
- [Eremothecium gossypii](#)
- [Gibberella zeae](#)
- [Kluyveromyces lactis](#)
- [Magnaporthe grisea](#)
- [Neurospora crassa](#)
- [Saccharomyces cerevisiae \(baker's yeast\)](#)
- [Schizosaccharomyces pombe \(fission yeast\)](#)
- [Ustilago maydis](#)
- [Yarrowia lipolytica](#)

NHGRI Current Topics in Genome Analysis 2006  
 Mining Genomic Sequence Data









NHGRI Current Topics in Genome Analysis 2006  
 Mining Genomic Sequence Data

**Gene**

NCBI Entrez Gene

Search | Gene for | Go | Clear

Display: Full Report Show: 5 Send to:

Ali: 1 Current Only: 1 Genes: 1 SNP GeneView: 1

1: ADAM2 - ADAM metallopeptidase domain 2 (fertilin beta) [Homo sapiens]

GenID: 2515 Primary source: HGNC:198

updated 17-Sep-2006

**Summary**

Official Symbol: ADAM2 and Name: ADAM metallopeptidase domain 2 (fertilin beta) provided by HUGO Gene Nomenclature Committee

See related: IPRD-03322, MIM-601533

Gene type: protein coding

Gene name: ADAM2

Gene description: ADAM metallopeptidase domain 2 (fertilin beta)

Gene symbols: ADAM2

Organism: *Homo sapiens*

Lineage: Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo

Gene aliases: FTNB; PH30; CRYN1; CRYN2; PH-30B

Summary: This gene encodes a member of the ADAM (a disintegrin and metalloprotease domain) family. Members of this family are membrane-anchored proteins similar in structure to snake venom disintegrins, and have been implicated in a variety of biological processes involving cell-cell and cell-matrix interactions, including fertilization, muscle development, and neurogenesis. This member is a subunit of an integral sperm membrane glycoprotein called fertilin, which plays an important role in sperm-egg interactions.

**Genomic regions, transcripts, and products**

(minus strand) RefSeq below

NC\_000008.9

39814906 39794111

NC\_011455.3

■ - coding region ■ - untranslated region

**Genomic context**

chromosome: 8; Location: 8p11.2

3947721 39952279

ADAM2

**Bibliography**

Gene References into Function (GeneRIF): Submit

**PubMed links**

**Interactions**

Description: ADAM2 Product Interactant Other Gene Complex Source Pubs

Done

**General gene information**

Markers: RH70674(e-PCR) (Links: UniSTS-33582), Alternate name: U52370, SHGC-111659(e-PCR) (Links: UniSTS-168466), ADAM2\_2713(e-PCR) (Links: UniSTS-462012)

**GeneOntology**

Provided by GOA

Function: integrin binding	Evidence: TAS	PubMed
Function: metalloendopeptidase activity	Evidence: IEA	
Function: protein binding	Evidence: IEA	
Process: cell adhesion	Evidence: IEA	
Process: fusion of sperm to egg plasma membrane	Evidence: TAS	PubMed
Process: proteolysis	Evidence: IEA	
Component: integral to plasma membrane	Evidence: TAS	PubMed
Component: membrane	Evidence: IEA	

**Homology:**  
 Mouse, Rat  
[Map Viewer](#)

**General protein information**

Names: ADAM metallopeptidase domain 2, fertilin beta; a disintegrin and metalloproteinase domain 2; a disintegrin and metalloproteinase domain 2 (fertilin beta)

NCBI Reference Sequences (RefSeq)

mRNA: NM\_021454  
 Source Sequence: RG719616.B1460477.U38805  
 Product: NP\_001455 ADAM metallopeptidase domain 2 proprotein  
 Conserved Domains (4) summary

pfaM01421: Repolysin/Repolysin (M12B) family zinc metalloprotease  
 Location: 170 - 375 Blast Score: 626  
 pfaM01421\_Pep\_M12B\_protein: Repolysin family propeptide  
 Location: 62 - 178 Blast Score: 362  
 smar00050: DISIN: Homologues of snake disintegrins ; Snake disintegrins inhibit the binding of ligands to integrin receptors  
 Location: 393 - 470 Blast Score: 281  
 smar00608: ACR; ADAM Cysteine-Rich Domain;  
 Location: 472 - 609 Blast Score: 375

**Related Sequences**

Nucleotide	Protein
Genomic: AC136365	None
Genomic: AP005902 (96106..113980, complement)	None
mRNA: AJ133005	CAB40813
mRNA: BC034957	AAH34957
mRNA: BC064547	AAH64547
mRNA: BG719616	None
mRNA: BJ460477	None

Done

**OMIM – A DISINTEGRIN AND METALLOPROTEINASE DOMAIN 2; ADAM2**

**Alternative titles; symbols**

FERTILIN, BETA; FTNB  
PH30

Gene map locus 8p11.2

**TEXT**

**DESCRIPTION**

The ADAMs (a disintegrin and metalloprotease domain) are a family of type I transmembrane glycoproteins that share homology with snake venom metalloprotease/disintegrins and sperm surface proteins. They are important in diverse biologic processes such as cell adhesion and proteolytic shedding of cell surface receptors. Structurally, ADAMs consist of a prodomain that blocks protease activity; a zinc-binding metalloprotease domain; disintegrin and cysteine-rich domains with adhesion activity; an epidermal growth factor (EGF 131530)-like domain with cell fusion activity; a transmembrane domain; and a phosphorylated cytoplasmic regulatory domain. For a review of the ADAM gene family, see Prmakoff and Myles (2000).

**CLONING**

Gupta et al. (1996) cloned human fertilin-beta (ADAM2). Using a guinea pig fertilin-beta cDNA as a probe to screen a human testis cDNA library, they obtained the 5'-prime end of the human fertilin-beta cDNA using RACE PCR. The complete human fertilin-beta cDNA contains an open reading frame of 2,199 bp and a 380-bp 3'-prime untranslated region. Gupta et al. (1996) compared the sequence of human fertilin-beta to related proteins and found metalloprotease, disintegrin, cysteine-rich, EGF-like repeat and transmembrane domains, a structural organization consistent with other members of the metalloprotease/disintegrin family. The amino acid sequence of the mature human fertilin-beta is 90% identical to monkey fertilin and 56 to 59% identical to mouse and guinea pig fertilin-betas. Gupta et al. (1996) performed Northern blot analysis of human fertilin-beta which detected a 3.2-kb transcript only in testis RNA, suggesting that human fertilin-beta may be specific to the testis.

Independently, Burkitt et al. (1997) cloned a human FTNB cDNA. They stated that the cDNA encodes a predicted 735-amino acid precursor protein from which the signal sequence (amino acids 1 to 16) and metalloprotease domain (amino acids 17 to 382) are cleaved during maturation.

**GENE FUNCTION**

Gupta et al. (1996) stated that most snake venom disintegrins contain the consensus integrin-binding sequence RGD. They noted that guinea pig, mouse, monkey, and human fertilin-betas contain tripeptide sequences TDE, QDE, FDE, and FEE at this location, respectively. These tripeptides are believed to mediate interaction with an integrin on the surface of the egg and thus mediate sperm/egg binding.

**HomoloGene**

**HomoloGene (hm)**

**Genes**

Genes identified as putative homologs of one another during the construction of HomoloGene.

- H. sapiens ADAM2
- P. troglodytes ADAM2
- C. familiaris LOC475578
- M. musculus Adam2
- R. norvegicus Adam2

**Proteins**

Proteins used in sequence comparisons and their conserved domain architectures.

NP_001455.3	735 aa
XP_519722.1	679 aa
XP_532795.2	881 aa
NP_033748.1	735 aa
NP_064462.1	739 aa

**Conserved Domains**

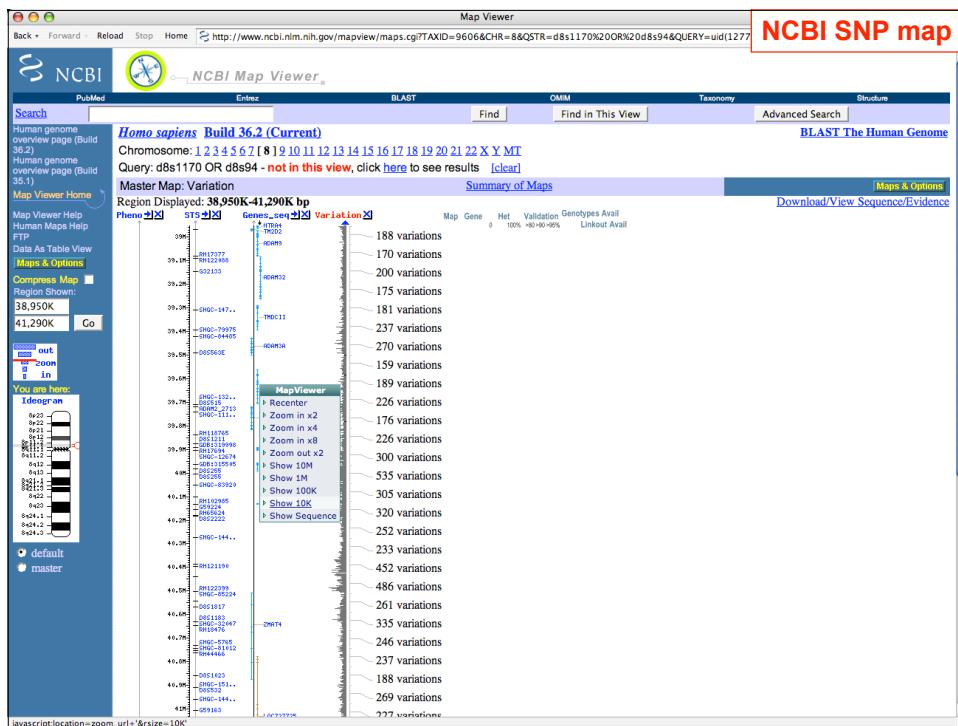
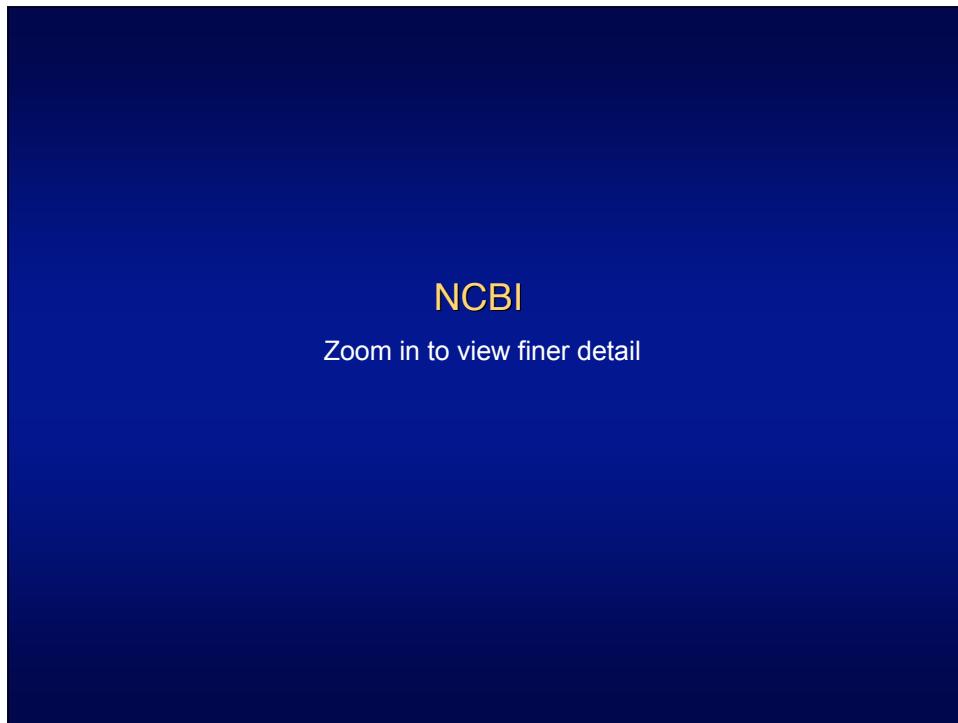
Conserved Domains from CDD found in protein sequences by *tblastn* searching.

- smart00050
- = DISIN-DISIN
- int-1
- = ACR-ACR
- pfdm1562
- = Pep\_M12B\_propep\_Pep\_M12B\_propep
- pfdm1421
- = Reprolysin, Reprolysin

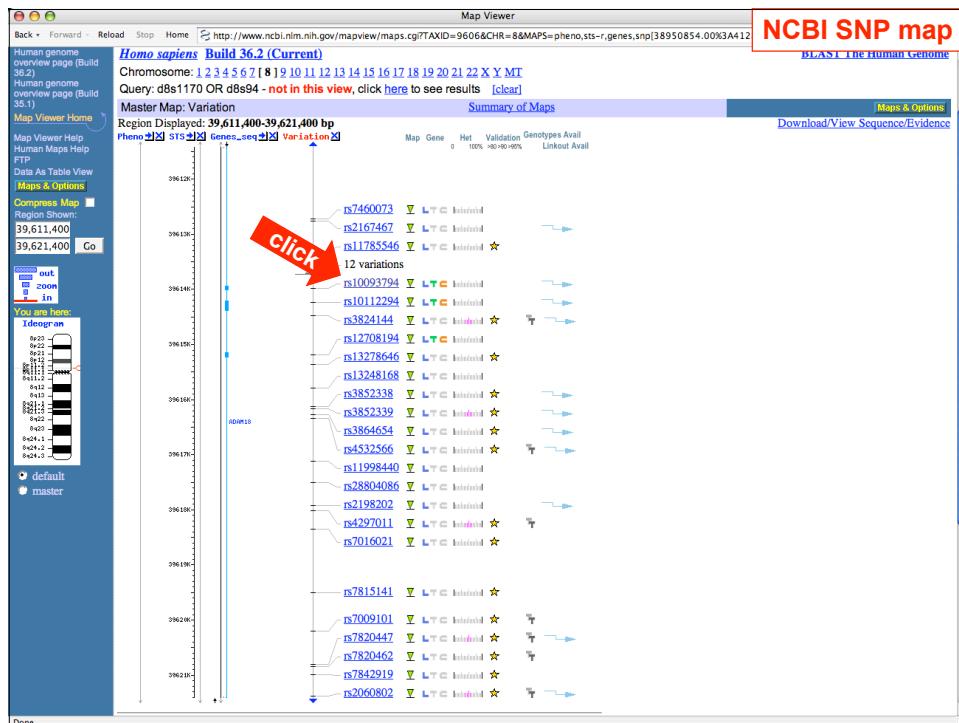
**Related Homology Resources**

Links to curated and computed homology information found in other databases.

MGI:1043894 Orthology group for M. musculus Adam2 includes H. sapiens ADAM2 and R. norvegicus Adam2.



NHGRI Current Topics in Genome Analysis 2006  
Mining Genomic Sequence Data



**NCBI Single Nucleotide Polymorphism**

Reference SNP (refSNP)

refSNP ID: rs10093794  
Organism: human (*Homo sapiens*)  
Molecule Type: Genomic  
Created/Updated in build: 119/126  
Map to Genome Build: 36.1

Allele: SNP: single nucleotide polymorphism  
Variation Class: Alleles: G/T  
Ancestral Allele: G

SNP Details are organized in the following sections: Submission, Fasta, Resource, GeneView, Map, Diversity, Validation, Linkout.

**Fasta sequence (Legend)**

```
>gnlHsSNPrs10093794[allelePos=501]totalLen=701taxid=9606[snpclass=1]alleles='G/T'!mol=Genomic!build=126
RCAAGGTTAC TGGCGCTTC AACACGTGAA GTTTCAGATT TTTCACATTT COAATTCTAT
GAAAGTTCTT TTAAATATAAT TTGGCGCTCA TTCTGGCCCA TGATTAACCTT AAATACCTTT
TATTGTTTA ATACGAAATA TACTGAAACA TGTGTTTTAT TTTTAAATGT ACCCTTTAAA
TGTAAATACT TTGTTCTTTA
```

**GeneView**

GeneView via analysis of contig annotation: ADAM18 a disintegrin and metalloproteinase domain 18

Click to see [all] [SNP] [has frequency] [double hit] [haplotype tagged] variations associated with this gene.

Group Label	Contig->mRNA	Gene Model (contig mRNA transcript) Color Legend			
reference	NT_007995>NM_014237	sv function			
Celera	NW_923907>NM_014237	sv function			
Group Label	Contig->mRNA->Protein	Contig position	mRNA orientation	mRNA pos	Function
reference	NT_007995>NM_014237>NP_055052	98152 7	forward	634	nonsynonymous T Phe (F) I 212
					contig reference G Val (V) I 212
Celera	NW_923907>NM_014237>NP_055052	27014487	forward	634	nonsynonymous T Phe (F) I 212
					contig reference G Val (V) I 212



**NCBI BLAST search**

BLAST Chicken Sequences.

Enter an accession, gi, or a sequence in FASTA format:  
inp\_001455

Or, choose a file to upload

Set subsequence: (optional)  
From: \_\_\_\_\_ To: \_\_\_\_\_

Database: genome (reference only) 40216 sequences

Program: BLASTN: Compare a protein sequence against a nucleotide database

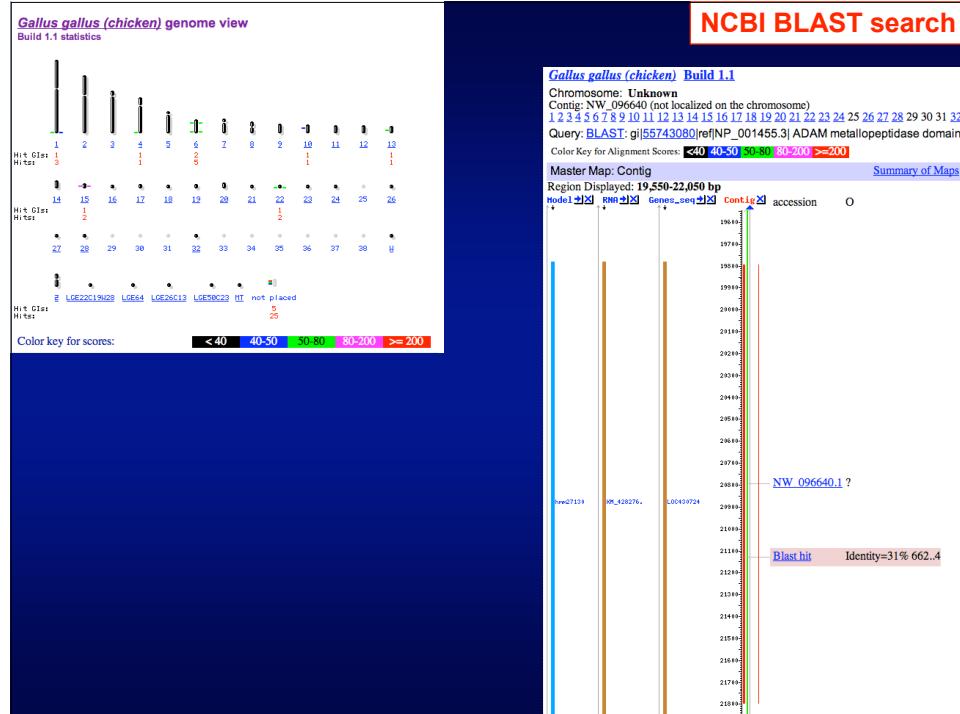
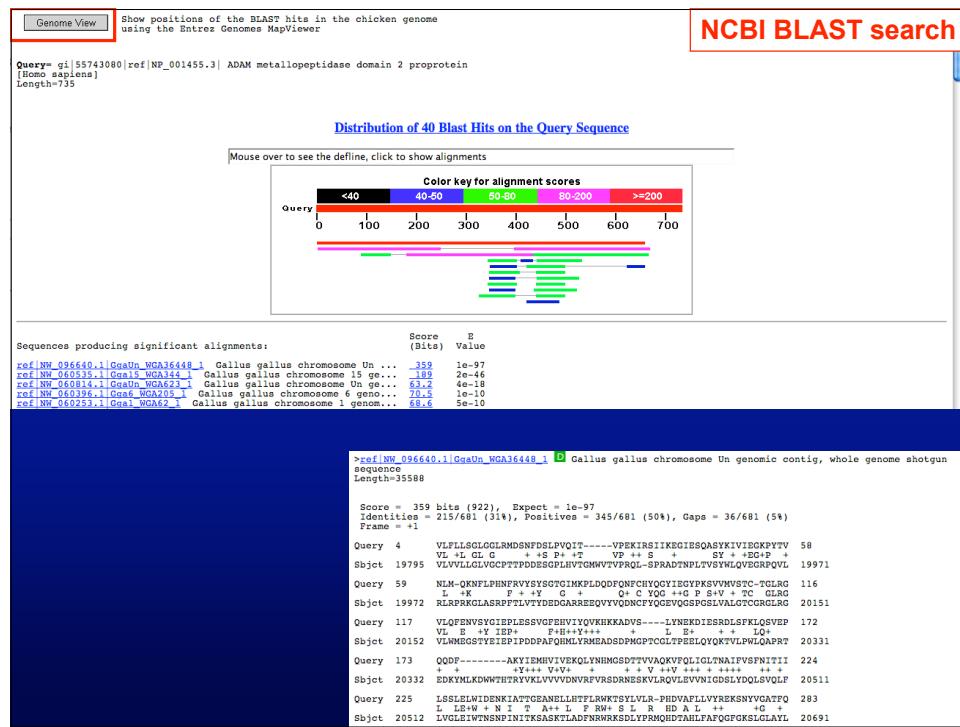
Optional parameters

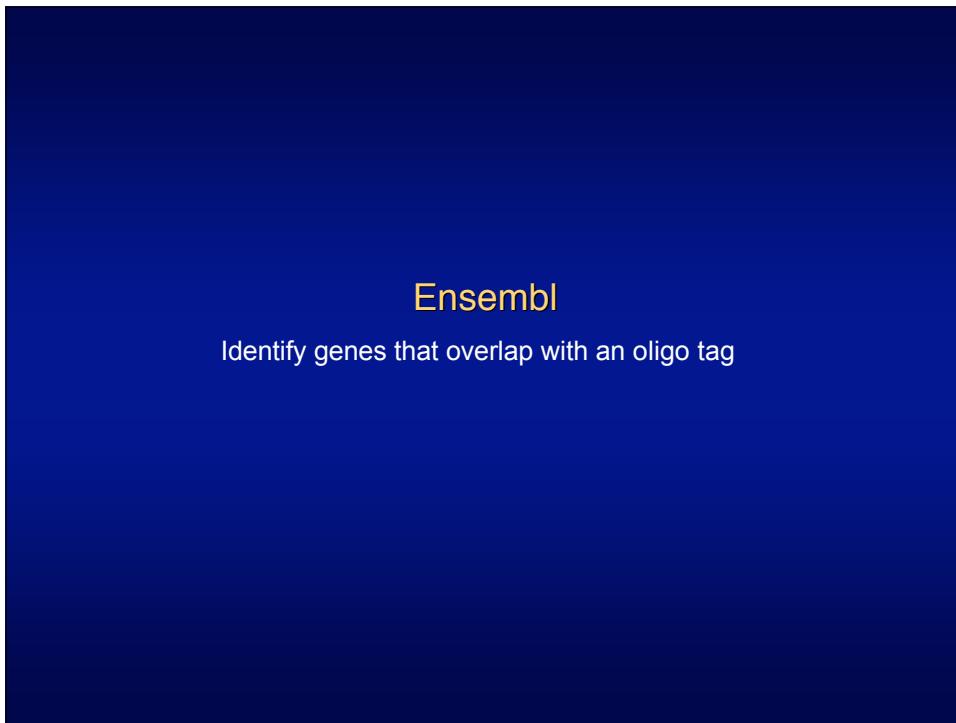
Expect: 0.01 | Filter: low complexity | Descriptions: 100 | Alignments: 100

Advanced options: \_\_\_\_\_

Begin Search | Clear Input |

Get the URL with preset values?





**click**

The screenshot shows the Ensembl Genome Browser homepage for release 40, dated August 2006. The page has a yellow header bar with the Ensembl logo and navigation links like 'Back', 'Forward', 'Reload', 'Stop', 'Home', and a search bar. Below the header is a large yellow sidebar containing links for 'Use Ensembl to...', 'Docs and downloads', and 'Other links'. A red arrow points to the 'Run a BLAST search' link in the 'Use Ensembl to...' section. The main content area is divided into several sections: 'What's New in Ensembl 40' (listing new low-coverage genomes, stickleback assembly, new species like Aedes aegypti, and new genebuilds for Macaque and Rat), 'Mammalian genomes' (listing Homo sapiens, Pan troglodytes, Macaca mulatta, Mus musculus, Rattus norvegicus, Oryctolagus cuniculus, Canis familiaris, Bos taurus, Sus scrofa, and others), and 'Other species' (listing Gallus gallus, Xenopus tropicalis, Danio rerio, Takifugu rubripes, Tetraodon nigroviridis, Gasterosteus aculeatus, Oryzias latipes, Ciona intestinalis, Ciona savignyi, Drosophila melanogaster, Anopheles gambiae, Aedes aegypti, Caenorhabditis elegans, and Saccharomyces cerevisiae). Logos for the Wellcome Trust, Sanger Institute, and EBI are visible at the bottom left.

**BlastView**

**Ensembl BLAST search**

Search all Ensembl: Anything | Go

**Ensembl Multi BlastView**

Ensembl release 40 - Aug 2006

**Use Ensembl to...**

- Run a BLAST search
- Search Ensembl
- Data mining [BioMart]
- Export data
- Download data

**Docs and downloads**

- Information
- What's New
- About Ensembl
- Ensembl data
- Software

**Other links**

- Home
- Sitemap
- Vega
- View previous release of page in Archive!
- Stable Archive link for this page
- Archive! sites
- Trace server

**Rattus norvegicus**  
**New genebuild**  
  
 Now in Ensembl!

**BLASTN SSANH2 TBLASTX**

**Search sensitivity:**  
 Optimise search parameters to find the following alignments

**About BlastView**

BlastView provides an integrated pipeline offering access to both BLAST and Ensembl databases. We would like to hear your suggestions in BlastView, especially regarding functionality that you would like to see provided in the future. Many thanks for your time! [\[Feedback\]](#)

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**BlastView**

**Ensembl BLAST search**

**Alignment Locations vs. Karyotype (click arrow to hide)**

**Alignment Locations vs. Query (click arrow to hide)**

**Alignment Summary (click arrow to hide)**

Select rows to include in table, and type of sort (Use the 'ctrl' key to select multiple)

Query	Subject	Chromosome	Supercontig	Clone	Contig	Chromosome	Stats	Sort By
Start	Name	Start	Name	Start	Name	Start	Off-Score	>Score
1A 1S1 1G 1C 1	21 + Chr.1s	54998166	54998166	0.09 +	Chr.1s	54998168	54998168	<Score
1A 1S1 1G 1C 1	17 + Chr.8	72205113	72205129	-	Chr.8	72205113	72205129	>Score

100% identity over 100% of the query length

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# NHGRI Current Topics in Genome Analysis 2006

## Mining Genomic Sequence Data

**Ensembl ContigView**

Ensembl v40: Homo sapiens Features on Chromosome 15 54996168-55000187

Back ▶ Forward ▶ Reload Stop Home http://www.ensembl.org/Homo\_sapiens/contigview?panel\_bottom=off;i=15%3A54996168-55000187;h=BL

e! Ensembl Human ContigView

Ensembl release 40 - Aug 2006

Chromosome 15  
54,996,168 - 55,000,187

- View of Chromosome 15
- Graphical view
- Graphical overview
- Export information about region
- Export sequence as FASTA
- Export EMBL file
- Export Gene info in region
- Export SNP info in region
- Export Vega info in region
- View alignment with ...
- View alongside ...
- View Syntenic regions ...
- View region in NCBI browser
- View region in UCSC browser

Use Ensembl to...

- Run a BLAST search
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- Download data

Docs and downloads

- Information
- What's New
- About Ensembl
- Ensembl data
- Software

Other links

- Home
- Sitemap
- Vega

Ensembl ContigView

Search □ Human □ Anything □ Go e.g. AL138722.15.1.44776, AL355340.17.1.112442 Help

Chromosome 15

Chr. 15 band

DNA(contigs)

Markers

Ensembl Genes

ncRNA Genes

EST Genes

Gene legend

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**Ensembl ContigView**

Ensembl v40: Homo sapiens Features on Chromosome 15 54996168-55000187

Back ▶ Forward ▶ Reload Stop Home http://www.ensembl.org/Homo\_sapiens/contigview?panel\_top=off;i=15%3A54996168-55000187;h=BL

e! Ensembl Human ContigView

Ensembl release 40 - Aug 2006

Chromosome 15  
54,996,168 - 55,000,187

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- Ensembl data
- Software

Other links

- Home
- Sitemap
- Vega

Ensembl ContigView

Search □ Human □ Anything □ Go e.g. AL138722.15.1.44776, AL355340.17.1.112442 Help

Chromosome 15

Overview

Detailed view

Features ▾ Comparative ▾ DAS Sources ▾ Repeats ▾ Decorations ▾ Export ▾ Image size ▾ Help ▾

Jump to region 15 : 54996168 - 55000187 Refresh Band: Refresh

<< 5MB < 2MB < 1MB < Window + Zoom Window 1MB 2MB > 5MB >

Chr. 15 Length Chr. 15 Length

EMBL mRNAs

Unigene

GenScan

EST trans.

Ensembl trans.

Blast hits

DNA(contigs)

Unigene

EMBL mRNAs

Length

Gene legend

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**Ensembl ContigView**

The screenshot shows the Ensembl v40 interface for Homo sapiens Features on Chromosome 15. The main panel is titled "Basepair view" and displays a sequence alignment. The alignment shows a forward strand from position 54,996,180 to 54,996,210. The sequence is color-coded by amino acid (e.g., R=red, K=blue, H=magenta, D=green, E=yellow, C=orange, P=pink, A=light blue, T=light green, S=light orange, N=light pink, M=light blue-grey). Above the sequence, there are tracks for Genscan, EST transcripts, and Ensembl transcripts. The bottom of the panel shows restriction enzyme sites for SphI, Ahal, BclI, Bss2, Bme, BssK, BssP, Eco1, Eco41, Eco51, Eco71, Eco81, and HinfI.

## Ensembl

Add features to the ContigView

# NHGRI Current Topics in Genome Analysis 2006

## Mining Genomic Sequence Data

**Ensembl ContigView**

The screenshot shows the Ensembl v40: Homo sapiens Features on Chromosome 15 54996168-55000187 page. The left sidebar contains a navigation menu with various links for viewing chromosomes, exporting data, and using Ensembl. A red arrow points to the 'select' button in the 'Detailed view' section of the sidebar. The main content area displays a genomic track for Chromosome 15, showing genes, transcripts, and other genomic features. The track includes a zoom function and a 'refresh' button. Below the track, there is a legend for gene and SNP tracks.

**Ensembl ContigView**

The screenshot shows the Ensembl v40: Homo sapiens Features on Chromosome 15 54996168-55000187 page. The left sidebar contains a navigation menu with various links for viewing chromosomes, exporting data, and using Ensembl. A red arrow points to the 'select' button in the 'Detailed view' section of the sidebar. The main content area displays a genomic track for Chromosome 15, showing genes, transcripts, and other genomic features. The track includes a zoom function and a 'refresh' button. Below the track, there is a legend for gene and SNP tracks. The bottom of the page includes copyright information and a note about available tracks.

The screenshot shows the Ensembl Archive Human ContigView interface for Chromosome 15. The URL is [http://feb2006.archive.ensembl.org/Homo\\_sapiens/contigview?panel\\_top=off&id=15&h=BLAST](http://feb2006.archive.ensembl.org/Homo_sapiens/contigview?panel_top=off&id=15&h=BLAST). The main panel displays a genomic track for Chr. 15, spanning from 54,996,168 to 55,000,187. The track includes various genomic features such as exons (red), introns (green), and other annotations (blue, purple). A search bar at the top right allows users to search for specific genes or regions. On the left, there is a sidebar with links for viewing the chromosome, detailed views, and various data sources like cDNAs, UniGene, and proteins.

## Ensembl

Get additional information about the gene, transcripts, and exons

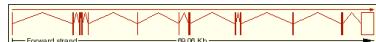


# NHGRI Current Topics in Genome Analysis 2006

## Mining Genomic Sequence Data

Ensembl GeneView

**Transcript ENST00000343827**

<b>Transcript</b>	<b>NP_996923.1</b> (RefSeq peptide) To view all Ensembl genes linked to the name <a href="#">click here</a> .
<b>Transcript information</b>	Exons: 13 Transcript length: 3,973 bps Protein length: 530 residues <a href="#">[Further Transcript Info]</a> <a href="#">[Exon Information]</a> <a href="#">[Protein Information]</a>
<b>Similarity Matches</b>	This Ensembl entry corresponds to the following database identifiers: RefSeq peptide: NP_996923.1 [Target ID: 96; Query ID: 100] [align] RefSeq DNA: NM_072601.1 [Target ID: 96; Query ID: 100] [align] UniProtKB/ProTMBL: Q27329_HUMAN [Target ID: 100; Query ID: 100] [align] Q9NQY1_HUMAN [Target ID: 7; Query ID: 100] [align] Q9NQY2_HUMAN [Target ID: 13; Query ID: 100] [align] Q9NQY4_HUMAN [Target ID: 4; Query ID: 100] [align] Q9NQY5_HUMAN [Target ID: 4; Query ID: 100] [align] Q9NQY6_HUMAN [Target ID: 9; Query ID: 100] [align] Q9NQY7_HUMAN [Target ID: 6; Query ID: 100] [align]
<b>EntrezGene:</b>	A_14_P120408 [Target ID: 1; Query ID: 100]
<b>Agilent CG-It:</b>	A_23_P151930 [Target ID: 1; Query ID: 100]
<b>Agilent Probe:</b>	A_24_P82142 [Target ID: 1; Query ID: 100]
<b>EMBL:</b>	AF221611 [align] AF221612 [align] AF221613 [align] AF221614 [align] AF221616 [align] AF221617 [align] BX537967 [align]
<b>IP:</b>	IP000384979.2 [Target ID: 96; Query ID: 100]
<b>Protein ID:</b>	AAFB2571.1 [align] AAFB2572.1 [align] AAFB2573.1 [align] AAFB2575.1 [align] AAFB2576.1 [align] AAFB2578.1 [align] CAD97931.1 [align]
<b>UniGene:</b>	Hs.385728 [Target ID: 9; Query ID: 95] Hs.388453 [Target ID: 12; Query ID: 99]
<b>Affymetrix Microarray Focus:</b>	208986_at
<b>Affymetrix Microarray HuGeneFL:</b>	208986_at
<b>Affymetrix Microarray U133:</b>	208986_at
<b>Affymetrix Microarray U95:</b>	238041_at
<b>Illumina:</b>	Hs.21704.0_S2_3p_at
	208986_at
	53348_at
	54311_at
<b>GO</b>	The following GO terms have been mapped to this entry via UniProt and/or RefSeq: <a href="#">GO:0005634 [nucleus]</a> <b>IEA</b> <a href="#">GO:0030528 [transcription regulator activity]</a> <b>IEA</b> <a href="#">GO:0045449 [regulation of transcription]</a> <b>IEA</b>
<b>InterPro</b>	IPR001092 Basic helix-loop-helix dimerisation region bHLH - <a href="#">[View other genes with this domain]</a>
<b>Protein Family</b>	ENSP00000000830 : TRANSCRIPTION FACTOR TRANSCRIPTION FACTOR This cluster contains 3 Ensembl gene member(s) in this species.
<b>Transcript structure</b>	
<b>Protein features</b>	

[http://www.ensembl.org/Homo\\_sapiens/geneview?gene=ENSG0000140262;db=core#ENST00000343827](http://www.ensembl.org/Homo_sapiens/geneview?gene=ENSG0000140262;db=core#ENST00000343827)

Ensembl ExonView

**Ensembl v40: Homo sapiens Exon Report for ENST00000343827;db=core**

<b>Archive sites</b>	<a href="#">Sanger</a> <a href="#">EBI</a>
<b>Intron 10</b>	ENSE00001126370 15 1 55,342,602 55,342,764 1 2 163
<b>Intron 10-11</b>	ENSE00001126365 15 1 55,342,765 55,352,519 2 1 9,755
<b>Intron 11-12</b>	ENSE00001103528 15 1 55,352,820 55,352,752 2 1 233
<b>Intron 12-13</b>	ENSE000011045489 15 1 55,362,889 55,365,646 9,182
<b>Intron 13</b>	ENSE000011045489 15 1 55,365,647 55,368,004 3,558

**3' downstream sequence**

**Supporting Evidence**

The supporting evidence below consists of the sequence matches on which the exon predictions were based and are sorted by alignment score.  
 There are a large number of supporting evidence hits for this transcript. Only the top ten 10 hits have been shown. [Click to view all 16 supporting evidence hits](#).

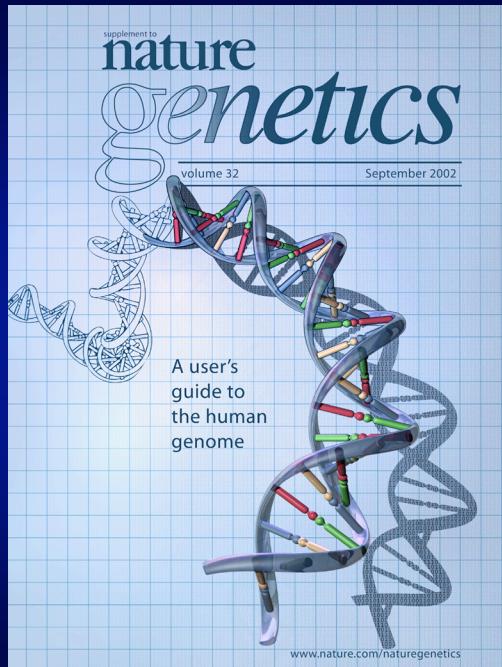
Score:  NO EVIDENCE

g|4637008|ref|NM\_207040.1| Homo sapiens transcription factor 12 (HSPF1), basic helix-loop-helix transcription factor...  
 g|4637008|ref|NM\_207037.1| Homo sapiens transcription factor 12 (HSPF1), basic helix-loop-helix transcription factor...  
 g|4637008|ref|NM\_207006.1| Homo sapiens transcription factor 12 (HSPF1), basic helix-loop-helix transcription factor...

[Done](#)

## Additional resources

- UCSC Human Genome Browser User Guide  
<http://genome.ucsc.edu/goldenPath/help/hgTracksHelp.html>
- NCBI Genomic Biology  
<http://www.ncbi.nih.gov/Genomes/>
- NCBI MapViewer Help  
<http://www.ncbi.nlm.nih.gov/mapview/static/MapViewerHelp.html>
- Ensembl Worked Example  
[http://www.ensembl.org/info/worked\\_example.pdf](http://www.ensembl.org/info/worked_example.pdf)



<http://www.nature.com/ng/supplements/>

## References

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